Programme Key:

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

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

WMS 2024 Full Programme

Version: 041024

Monday 7th October 2024

08:30-19:00	**Pre-Congress Teaching Course** Congress Venue, Club D+E (separate registration required)

Tuesday 8th October 2024

08:00-11:00	**Pre-Congress Teaching Course** Congress Venue, Club D+E (separate registration required)	
11:00-16:00	**WMS Executive Board Meeting** South Boardroom 1	
14:30-18:00	**Registration and poster set up** Congress Venue, Second Floor Foyers	
14:30-16:00	**Meet the Exhibitors** Congress Venue, Second Floor Foyers (Catering provided)	
16:30-17:30	**Industry Symposium 1** South Hall 1	**Industry Symposium 2** South Hall 2 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium
18:00-18:45	**Opening Ceremony** Congress Hall (separate registration required) *Moderators: Jana Haberlová, University Hospital Motol, Czechia & Radim Mazanec, Charles University, Czechia*	
18:45-21:00	**Networking Reception** Congress Venue, Panorama Hall and Zoom Room, First Floor (separate registration required)	

Wednesday 9th October 2024

06:30-19:30	**Congress desk open**	
07:45-08:45	**Industry Symposium 3** South Hall 1 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium	**Industry Symposium 4** South Hall 2 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium
09:00-09:15	**Congress Welcome - Message from the President** Congress Hall	
09:15-10:45	**Topic 1: Acquired Muscle Disorders** Congress Hall *Moderators: Suur Biliciler, Uthealth Science Center Houston, Mcgovern Medical School, USA & Heřman Mann, Institute of Rheumatology Prague, Czechia*	

09:15-09:45	**01INV** Acquired muscle disorders: Introduction to CART cells and Tregs ***Villalta, A***[1,2,3] [1]*Physiology and Biophysics,* [2]*Muscle Biology and Disease Research Center,* [3]*Institute for Immunology, University of California Irvine*
09:45-10:15	**02INV** Current and prospective uses of CAR and CAAR T-cell therapies in muscle disorders ***Mozaffar, T***[1] [1]*University of California Irvine*
10:15-10:30	**01O** Response to rozanolixizumab across treatment cycles in patients with generalised Myasthenia Gravis: A post hoc analysis ***Vissing J***[1], *Grosskreutz J*[2], *Habib A*[3], *Mahuwala Z*[4], *Mantegazza R*[5], *Pascuzzi R*[6], ***Sacconi S***[7], *Vu T*[8], *Beau Lejdstrom R*[9], *Greve B*[10], *Grimson F*[11], *Tarancón T*[12], *Bril V*[13] [1]*University of Copenhagen,* [2]*University of Lübeck,* [3]*University of California,* [4]*Department of Neuromuscular Medicine and Clinical Neurophysiology, University of Kentucky, USA,* [5]*Istituto Nazionale Neurologico Carlo Besta,* [6]*Indiana University School of Medicine,* [7]*Université Côte d'Azur, 8University of South Florida Morsani College of Medicine,* [9,10,11,12]*UCB Pharma,* [13]*University Health Network Canada*
10:30-10:45	**02O** Characterization of a mouse model for Jo-1, PL-7 and PL-12 associated Anti-synthetase syndrome ***Bachir D***[1], *Preusse C*[2], *Lichtenberg S*[1], *Koch-Hölsken K*[1], *Umathum V*[3,4], *Herrmann A*[1], *Schaenzer A*[3], *Meuth S*[1], *Stenzel W*[2], *Ruck T*[1] [1]*Heinrich Heine University,* [2]*Charité-Universitätsmedizin Berlin,* [3]*Justus Liebig University,* [4]*German Armed Force Hospital*
10:45-11:15	**Morning refreshments, exhibition and posters** Congress Hall Foyer, Forum Hall and Forum Foyer
10:45-11:15	**Myology Café - Meet the WMS EDI Committee** Congress Hall Foyer
11:15-13:15	**Topic 1: Acquired Muscle Disorders** Congress Hall *Moderators: Ichizo Nishino, National Institute of Neuroscience, Japan & Corinna Preusse, Charité-universitätsmedizin Germany*
11:15-11:45	**03INV** Idiopathic inflammatory myopathies: current state of the field, new insights and treatment ***Vencovský J***[1], *Mann H*[1] [1]*Institute of Rheumatology, Czechia*
11:45-12:15	**04INV** Acquired muscle disease - a paediatric perspective ***Ramdas S***[1] [1]*Oxford University Hospitals NHS foundation Trust, Oxford, UK,* [2]*University of Oxford, Oxford, UK*
12:15-12:30	**03O** Identification of Class I HLA genetic predispositions and prediction of autoantigenic epitopes in dermatomyositis patients of Indian origin ***Jassal B***[1], *Deepak R*[2], *Venugopalan Y V* [3], *Suri V*[1], *Sharma M*[1] [1]*Neuropathology Lab, All India Institute of Medical Sciences,* [2]*Department of Transplant Immunology and Immunogenetics, All India Institute of Medical Sciences,* [3]*Department of Neurology, All India Institute of Medical Sciences*
12:30-12:45	**04O** In-silico Interactomics as a way to elucidate Inclusion Body Myositis ***De Los Reyes F***[1], *Hayashi S*[1], *Noguchi S*[1], *Nishino I*[1] [1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry*
12:45-13:00	**05O** Unravelling the role of early mitochondrial dysfunction in Inclusion Body Myositis: a chicken or egg dilemma reloaded ***Kleefeld F***[1], *Cross E*[2], *Lagos D*[2], *Preusse C*[1], *Roos A*[3,4,5,6], *Horvath R*[2], *Stenzel W*[1] [1]*Charité - Universitätsmedizin Berlin,* [2]*University of Cambridge,* [3]*Department of Neurology, University Hospital Düsseldorf, Heinrich Heine University,* [4]*Brain and Mind Research Institute, Children's Hospital of Eastern Ontario Research Institute,* [5]*Leibniz-Institut für Analytische Wissenschaften-ISAS e.V,* [6]*Department of Pediatric Neurology, Centre for Neuromuscular Disorders, Centre for Translational Neuro- and Behavioural Sciences, University Duisburg-Essen*
13:00-13:15	**06O** NLRP3 inflammasome activation and altered mitophagy are key pathways in Inclusion Body Myositis. ***Naddaf E***[1], *Nguyen T*[1], *Watzlawik J*[2], *Gao H*[3], *Hou X*[2], *Fiesel F*[2,4], *Mandrekar J*[1,5], *Kokesh E*[1], *Harmsen W*[5], *Lanza I*[6], *Springer W*[2,4], *Trushina E*[1,3] [1]*Department of Neurology, Mayo Clinic,* [2]*Department of Neuroscience, Mayo Clinic,* [3]*Department of Molecular Pharmacology and Experimental Therapeutics, Mayo Clinic,* [4]*Neuroscience PhD Program, Mayo Clinic Graduate School of Biomedical Sciences,* [5]*Department of Quantitative Health Sciences, Mayo Clinic,* [6]*Division of Endocrinology and Metabolism, Mayo Clinic*

13:15-14:30	**Lunch, exhibition and posters** Congress Hall Foyer, Forum Hall and Forum Foyer
13:15-14:30	**Meet the Experts Lunch** Zoom and Panorama Rooms, 1st Floor (separate registration required and lunch is provided)
14:30-15:30	**Poster Session 1** Forum Hall (lunch is provided)

113P-146P, 147VP: SMA Clinical

113P Quality of life and participation of adults with spinal muscular atrophy: QOLSMA
Ribault S[1], *Rippert P*[1], *Lopinet T*[1], *Le Goff L*[1], *Barrière A*[1], *Morard M*[1], *Theuriet J*[1], *Pegat A*[1], *Boyer F*[2], *Vuillerot C*[1]
[1]*Hospices Civils De Lyon,* [2]*CHU de Reims*

114P Epidemiology and therapeutic outcomes of patients with Spinal Muscular Atrophy: results from a 12-year real-world study based on the French National Healthcare database (SNDS)
Quijano-Roy S[1], *Bourget I*[2], *Lot A*[1], *Desguerre I*[3], *Urtizberea J*[4], *de Chasteigner A*[5], *Leiba G*[5], *Affinito S*[5], *Panes A*[6], *Denis H*[6], *Schmidt A*[6]
[1]*APHP Raymond Poincare Hospital,* [2]*Gustave Roussy Hospital,* [3]*APHP Necker Enfants Hospital,* [4]*GH Pitie Salpetriere,* [5]*Novartis GT France SAS,* [6]*HEVA*

115P MRI of whole-body muscles and tongue of patients with Spinal Muscular Atrophy
Froelich S[1], *Receveur N*[1], *Scharff Poulsen N*[1], *Espe Hansen A*[2], *Vissing J*[1]
[1]*Copenhagen Neuromuscular Center, (CNMC), Department of Neurology, Rigshospitalet,* [2]*Department of Radiology, Rigshospitalet*

116P Exploring neurobehavioral disorders in type 1 and presymptomatic patients with Spinal Muscular Atrophy
Buchignani B[1,2], *Coratti G*[2,3], *Cutrì C*[3], *Palermo C*[2], *Leone D*[2], *Antonaci L*[2], *Pera M*[2,3], *Pane M*[2,3], *Mercuri E*[2,3]
[1]*Università Di Pisa,* [2]*Centro Clinico Nemo Pediatrico,* [3]*Università Cattolica del Sacro Cuore*

117P Prevalence of neurodevelopmental anomalies in patients with Spinal Muscular Atrophy Type I registered in CUIDAME
Alvarez Molinero M[1], *Gómez Andrés D*[1], *Garcia Uzquiano R*[2], *Expósito Escudero J*[2], *Costa Comellas L*[1], *Garcia Romero M*[3], *Martinez Salcedo E*[4], *Nungo Garzón C*[5], *Grimalt Calatayud M*[6], *Lopez Lobato M*[7], *Munell Casadesús F*[1], *Fernández Ramos J*[8], *Calzada, Garcia-Mora C*[9], *Gómez Martín H*[10], *Calvo R*[11], *Toledo Bravo de Laguna L*[12], *González Barrios D*[13], *Navarrio Abia V*[14], *Fernández Garcia M*[3], *CUIDAME Investigators Group*[15]
[1]*Vall Hebron University Hospital,* [2]*Sant Joan de Deu Hospital,* [3]*La Paz University Hospital,* [4]*Virgen de la Arrixaca University Hospital,* [5]*La Fe Valencia Hospital,* [6]*Son Espases University Hospital,* [7]*Virgen del Rocio Hospital,* [8]*Reina Sofía Hospital,* [9]*Toledo University Hospital,* [10]*Salamanca University Hospital,* [11]*Regional University Hospital of Malaga,* [12]*Materno-Infantil Hospital of Las Palmas de Gran Canaria,* [13]*Nuestra Señora de Candelaria University Hospital,* [14]*University Hospital of Burgos,* [15]*Cuidame Investigators Group*

118P Characterization of patients with Type 1 Spinal Muscular Atrophy in advanced disease state treated with Nusinersen
Balkenhol J[1], *Beytia M*[1], *Jofre J*[2], *Suarez B*[2], *Hervias C*[3], *Calcagno G*[3], *Castiglioni C*[2]
[1]*Pontificia Universidad Católica De Chile, División Pediatrìa, Unidad Neurología Pediátrica,* [2]*Clinica Meds, Neurology Service,* [3]*Clinica Meds, Neurokinesiology Service*

119P Evaluation of physical function before and after medical treatment in patients with Spinal Muscular Atrophy aged 11-25 years
Werlauff U[1], *Busk L*[1], *Drivsholm P*[1], *Heiden B*[1], *Iversen R*[1], *Laustsen H*[1], *Mahoney A*[1], *Olesen L*[1], *Handberg C*[1,2]
[1]*The Danish National Rehabilitation Centre for Neuromuscular Diseases,* [2]*Department of Public Health, Faculty of Health, Aarhus University*

120P Diagnostic delay in Korean adults with Spinal Muscular Atrophy
Kim S[1], *Choi Y*[1], *Cho J*[2], *Choi Y*[1], *Park H*[1]
[1]*Department Of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine,* [2]*Department of Neurology, National Health Insurance Service Ilsan Hospital*

121P Insulin resistance and liver fibrosis and - steatosis in adult patients with Spinal Muscular Atrophy
Receveur N[1], *Frølich S*[1], *Scharff Poulsen N*[1], *Ewertsen C*[2], *Espe Hansen A*[2], *Vissing J*[1]
[1]*Copenhagen Neuromuscular Center, Department of Neurology, Rigshospitalet,* [2]*Department of Radiology, Rigshospitalet*

122P A retrospective review of changes in upper limb function following spinal surgery in Spinal Muscular Atrophy
Martineau O[1,2], *Milev E*[1,2], *Main M*[1], *Scoto M*[1,2], *Muntoni F*[1,2], *Baranello G*[1,2]
[1]*Great Ormond Street Hospital,* [2]*University College London*

123P The difficult path to diagnosis of the patient with spinal muscular atrophy

Bolano Diaz C[1,2], Morosini M[1], Chloca F[1], Mesa L[1], Jauregui A[1], Pirra L[1], Vazquez G[1], Flores D[1], Dubrovsky A[1]

[1]Favaloro Foundation - Neurosciences Institut, [2]John Walton Muscular Dystrophy Research Centre

124P Hidden in plain sight: genome reanalysis to identify an intragenic novel variant in the SMN locus in a patient with an undiagnosed lower motor neuron disease

Haliloğlu G[1], Donkervoort S[2], Weisburd B[3,4], Öz Yıldız S[1], Ceylaner S[5], Pais L[3,6], O'Donnell-Luria A[3,6], Bönnemann C[2]

[1]Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, [2]Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, [3]Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, [4]Center for Genomic Medicine, Massachusetts General Hospital, [5]Intergen Genetic Diagnosis and Research Center, [6]Division of Genetics and Genomics, Boston Children's Hospital, Harvard Medical School

125P A review of the management and outcomes of children with SMA in the West Midlands, UK during 2017-2022

Willis T[1,2], Kulshrestha R[1], Alhaswani Z[3], Parasuraman D[3], Kuiper J[1], Jones J[1], Perry C[1]

[1]Robert Jones and Agnes Hunt hospital, [2]University of Chester Medical School, [3]University Hospitals Birmingham

126P Retrospective assessment of feeding and nutrition after 2 years of risdiplam treatment in children with Type 1 SMA using a novel scale

Baranello G[1,2], Conway E[1], Li Y[3], Gorni K[4]

[1]Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, UCL Great Ormond Street Institute of Child Health & Great Ormond Street Hospital Trust, [2]Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, [3]Pharma Development, Data Sciences, F. Hoffmann-La Roche Ltd, [4]PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd

127P UK real-world longitudinal data collection and analysis in adult SMA: theAdult SMA REACH Data collection study

Karkkainen E[1,2], Murphy L[1,2], Muni Lofra R[1,2], Segovia S[1,2], Page J[1,2], Verdu-Diaz J[1], Michell-Sodhi J[1,2], Alvarez G[1,2], Marini-Bettolo C[1,2]

[1]The John Walton Muscular Dystrophy Research Centre, [2]Newcastle University and Newcastle Hospitals NHS Foundation Trust

128P Treatment with gene therapy in patients with spinal muscular atrophy. Current experience in CuidAME registry

Exposito Escudero J[1], Costa Comellas L[2], García Romero M[3], Toledo Bravo De Laguna L[4], Gomez Martin H[5], Calvo Medina R[6], Navarro Abia V[7], Ñungo Garzón C[8], Martinez Gonzalez M[9], Martínez Salcedo E[10], Grimalt Calatayud M[11], Martínez González M[12], Álvarez Molinero M[2], Fernández García M[3], Puig Ram C[1], Garcia Uzquiano R[1], Hervás D[8], Nascimento Osorio A[1], CUIDAME investigators Group[13]

[1]Hospital Sant Joan De Deu, [2]Hospital Vall d'Hebron, [3]Hospital Universitario La Paz, [4]Hospital Materno-Infantil de Canarias, [5]Complejo Asistencial Universitario de Salamanca, [6]Hospital Regional Universitario de Malaga, [7]Hospital Universitario de Burgos, 8Hospital Universitario y Politecnico la Fe, [9]Hospital Universitario de Cruces, [10]Hospital Clinico Universitario Virgen de la Arrixaca, [11]Hospital Son Espases, [12]Complejo Universitario Hospital de Albacete, [13]CUIDAME investigators Group

129P Newborn screening for Spinal Muscular Atrophy in Poland – a summary of 3-year experience

Gos M[2], Wasiluk J[2], Landowska A[2], Jurzyk M[2], Wawer W[2], Kubiszyn P[2], Wieczorek J[2], Kordowska O[2], Nosarieva L[2], Durda K[3], Fraczyk M[2], Jedrzejowska M[1], Ołtarzewski M[2]

[1]Medical University of Warsaw, [2]Institute of Mother and Child, [3]Pomeranian Medical University

130P Real-life outcome data on Risdiplam therapy for Spinal Muscular Atrophy

Lavi R[1,2], Sagi L[1,2], Katzenellenbogen S[1], Shtamler A[1], Weizman A[1], Opincariu I[1,3], Fattal-Valevski A[1,2]

[1]Sourasky Medical Center, [2]Faculty of Medicine Tel Aviv University, [3]Schonbrunn Academic School of Nursing, Sourasky Medical Center

131P Social communication abilities in treated children with Spinal Muscular Atrophy type 1 (SMA1): a cross-sectional study from two tertiary neuromuscular centres

Brusa C[1,2], Buchignani B[3,4], Cutri C[5], Coratti G[3,5], Weststrate H[1,2], Clark E[2], Johnson E[2], Barritt E[2], Antonaci L[3], Leone D[3], Palermo C[3], Cornell N[1,2], Munot P[2], Manzur A[2], Scoto M[1,2], Pane M[3,5], Mercuri E[3,5], Muntoni F[1,2], Baranello G[1,2]

[1]UCL - Great Ormond Street Institute of Child Health, [2]Great Ormond Street Hospital for Children, [3]Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli, IRCCS, Rome, [4]University of Pisa, 5Pediatric Neurology, Università Cattolica del Sacro Cuore

132P Early-onset motor neuropathies and novel genetic causes: a Chilean series

Castiglioni Toledo C[1]

[1]Clinica Meds-Inrpac

133P Description of UK SMA cohorts since the introduction of disease modifying therapies

Muni Lofra R[1,2], *Rowher A*[3], *Muntoni F*[3], *Marini-Bettolo C*[1,2], *SMAREACH UK Sterring Group, Adult SMAREACH Steering Group*

[1]*John Walton Muscular Dystrophy Research Centre-Newcastle University,* [2]*Newcastle Hospitals NHS Foundation Trust,* [3]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health*

134P Beyond the "walker" label: functional diversity and disease impact in spinal muscular atrophy patients with walking ability

Cattinari M[1], *Hervás D*[2], *de Lemus M*[1,3,4], *Tizzano E*[1,5]

[1]*Fundame, Madrid, Spain,* [2]*Department of Applied Statistics and Operations Research and Quality, Universitat Politècnica de València,* [3]*SMA Europe Germany,* [4]*Committee of Advanced Therapies at the European Medicines Agency,* [5]*Department of Clinical and Molecular Genetics and Rare Diseases Unit and Medicine, Genetics Group, VHIR, Hospital Vall d'Hebron*

135P Use of electrophysiologic data and physical therapy assessments in SMA monitoring

Wright M[1,2], *Yang M*[1,2], *Moore Burk M*[2], *Kelley C*[2], *Stratton A*[1,2], *Apkon S*[1,2], *Gibbons M*[1,2], *Browning K*[1,2], *Kupfer O*[1,2], *Watne L*[2], *Foster H*[2], *Parsons J*[1,2]

[1]*University of Colorado School of Medicine,* [2]*Children's Hospital Colorado*

136P Medical treatment of children with spinal muscular atrophy - An investigation of parents' experiences of hopes, worries and need for rehabilitation for their child

Handberg C[1,2], *Werlauff U*[1], *Drivsholm P*[1,2], *Lorenzen S*[1], *Mahoney A*[1]

[1]*National Rehabilitation Center for Neuromuscular Diseases,* [2]*Department of Public Health, Faculty of Health, Aarhus University*

137P Mortality in Spinal Muscular Atrophy in the era of disease-modifying therapies

Finnegan R[1], *Rohwer A*[1], *Scoto M*[1,2], *Main M*[1], *Baranello G*[1,2], *Manzur A*[1], *Muntoni F*[1,2], *Munot P*[1]

[1]*Dubowitz Neuromuscular Centre, Great Ormond Street Hospital for Children,* [2]*NIHR Great Ormond Street Hospital Biomedical Research Centre*

138P Long-term motor responses to disease-modifying therapies in Spinal Muscular Atrophy (SMA) adults: a prospective study

Duong T[1], *Tang W*[1], *Gu B*[1], *Dunaway Young S*[1], *Corratti G*[2], *Pasternak A*[4], *Glanzman A, de Monts C*[1], *Rodriguez-Torrres R*[3], *Maczek E*[4], *Martens B*[5], *Salvator S*[1], *Smith S*[1], *Yatsu L*[1], *Hailu R*[1], *Park T*[1], *He Z*[1], *Mercuri E*[2], *Day J*[1]

[1]*Stanford University,* [2]*Catholic University of Sacred Heart,* [3]*Columbia University,* [4]*Boston Children's Hospital,* [5]*University of Rochester,* [6]*Childrens Hospital of Philadelphia,*

139P Treatment effects on ambulation loss in Spinal Muscular Atrophy Type III: insights from the Italian ISMAC registry

Coratti G[1], *D'Amico A*[2], *Sansone V*[3], *Masson R*[4], *Maggi L*[5], *Mongini T*[6], *Pegoraro E*[7], *Pini A*[8], *Vacchiano V*[9], *Coccia M*[10], *Filosto M*[11], *Ticci C*[12], *D'Errico E*[13], *Nigro V*[14], *Bruno C*[15], *Messina S*[16], *Pera M*[1], *Pane M*[1], *Mercuri E*[1]

[1]*Child Neurology Unit E Centro Nemo, IRCCS Fondazione Policlinico Gemelli, Università Cattolica del Sacro Cuore,* [2]*Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesú Children's Hospital IRCCS,* [3]*Centro Clinico NeMo,* [4]*Pediatric Neurology and Myopathology Units, Neurological Institute "Carlo Besta",* [5]*Neurology IV, Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta,* [6]*Neuromuscular Center, "Rita Levi Montalcini" Department of Neuroscience, University of Turin,* [7]*Department of Neurosciences DNS, University of Padova,* [8]*Department of Biomedical and Neuromotor Sciences, IRCCS Istituto delle Scienze Neurologiche di Bologna, University of Bologna,* [9]*UOC Clinica Neurologica, IRCCS Istituto delle Scienze Neurologiche di Bologna,* [10]*Centro NeMO Ancona,* [11]*Department of Clinical and Experimental Sciences, University of Brescia; NeMO-Brescia Clinical center for Neuromuscular Diseases,* [12]*SOC Malattie Metaboliche e Muscolari Ereditarie, Meyer Children's Hospital IRCCS,* [13]*Neurology Unit, Azienda Ospedaliero-Universitaria, Policlinico of Bari,* [14]*Medical Genetics and Cardiomyology, Department of Precision Medicine, University of Campania "Luigi Vanvitelli",* [15]*Center of Translational and Experimental Myology and Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, IRCCS Istituto Giannina Gaslini and University of Genoa,* [16]*Department of Clinical and Experimental Medicine, University of Messina*

140P Need for tube feeding in SMA type I patients treated with disease modifying therapies: bulbar function before treatment matters

Coratti G[1], *Pane M*[1], *Stanca G*[1], *D'Amico A*[2], *Sansone V*[3], *Berti B*[1], *Fanelli L*[1], *Albamonte E*[3], *Ausili C*[1], *Cerchiari A*[4], *Catteruccia M*[2], *De Sanctis R*[1], *Leone D*[1], *Palermo C*[1], *Buchignani B*[1], *Tosi M*[2], *Pera M*[1], *Bertini E*[2], *Mercuri E*[1]

[1]*Child Neurology Unit e Centro Nemo, IRCCS Fondazione Policlinico Gemelli, Università Cattolica del Sacro Cuore,* [2]*Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesú Children's Hospital IRCCS,* [3]*Centro Clinico NeMo,* [4]*Feeding and Swallowing Services Unit, Bambino Gesù Children's Hospital IRCCS*

141P Spinal Muscular Atrophy is also a disorder of spermatogenesis

Magot, A[1], *Reignier, A*[2], *Binois, O*[3], *Vuillerot,C*[4,5], *Yann, P*[1] *and the Fermasi Study Group*

[1]*Centre de Référence des Maladies Neuromusculaires AOC, CHU de Nantes, Filnemus, Euro-NMD, Nantes,* [2]*Service de Médecine et Biologie de la Reproduction, gynécologie médicale, CHU de Nantes,* [3]*Service de Biologie de la Reproduction - CECOS, Hôpital Antoine Béclère, AP-HP, Université Paris Saclay,* [4]*Centre de Référence PACA Réunion Rhône Alpes, Hospices Civils de Lyon, Hôpital Femme-Mère-Enfant, L'Escale, Service de Médecine Physique et de Réadaptation Pédiatrique, Bron,* [5]*NeuroMyogen Institute, CNRS UMR 5310 - INSERM U1217, University of Lyon*

142P A preliminary machine learning retrospective observational study to predict treatment response to nusinersen in non-sitter Spinal Muscular Atrophy

Stimpson G[1,2,6], *O'Reilly E*[1,2], *Coratti G*[3,4], *Ridout D*[5], *Chakraborti T*[6,7], *Mitra R*[6,8], *Mercuri E*[3,4], *Scoto M*[3,4], *Muntoni F*[3,4], *Baranello G*[3,4]

[1]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health,* [2]*National Institute for Health Research Great Ormond Street Hospital Biomedical Research Centre,* [3]*Pediatric Neurology Unit, Catholic University,* [4]*Centro Clinico Nemo, U.O.C. Neuropsichiatria Infantile Fondazione Policlinico Universitario Agostino Gemelli IRCCS,* [5]*Population, Policy & Practice Department, UCL Great Ormond Street Institute of Child Health,* [6]*Turing-Roche Strategic Partnership, Alan Turing Institute,* [7]*Dept of Medical Physics and Biomedical Engineering and the Dept of Pathology, UCL Cancer Institute,* [8]*Department of Statistical Science, University College London*

143P Swallowing physiology and function in untreated patients with Spinal Muscular Atrophy type 1: establishing natural history reference values

McGrattan K[1], *Graham R*[2], *Miles A*[3], *Allen J*[3], *Hofelich Mohr A*[1], *Rao V*[4], *Alfano L*[5], *Smith L*[11], *Brandesma J*[6], *Leon Astudillo C*[7], *Levy D*[8], *Tang W*[10], *Brown A*[13], *Spoden A*[1], *Schenck G*[9], *Darras B*[2]

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144P Gene therapy for Spinal Muscular Atrophy amid the dengue outbreak in Brazil: a case report

Graca F[1], *C França Jr M*[1]

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145P Efficacy of SMA NBS: 4-year comparative study with control group

Dangouloff T[1], *De Waele L*[2], *Beysen D*[3], *Smeets N*[4], *Vanlander A*[5], *Benmhammed N*[1], *Tychon C*[1], *Daron A*[1], *Deconinck N*[6], *Dontaine P*[6], *Servais L*[1,7]

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146P Investigation of bone health in a large cohort of naïve SMA patients

Panicucci C[1], *Brolatti N*[1], *Casalini S*[1], *Coratti G*[2], *Pedemonte M*[1], *Ricci F*[3], *Mongini T*[4], *Sansone V*[5], *Filosto M*[6], *Bello L*[7], *Pegoraro E*[7], *Bruno I*[8], *Verriello L*[9], *Ricci G*[10], *D'Amico A*[11], *Mercuri E*[2], *Maghnie M*[1], *Di Iorgi N*[1], *Bruno C*[1], *working group ITASMAC*[12]

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147VP Efficacy and safety of Nusinersen in the treatment of Spinal Muscular Atrophy in adolescents and adults

Ningning W[1,4,5], *Hu Y*[2], *Yu L*[3], *Zhu W*[1,4,5]

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204P-231P, 232VP-233VP: Clinical trials, access to health care and outcome measures

204P A phase 1 study of antisense oligonucleotide NS-035 in patients with Fukuyama Congenital Muscular Dystrophy

Fujino G[1], Kitamura A[1], Takahashi A[1], Maeda M[1], Kubota A[1], Tokuyama Y[2], Wada I[2], Kobayashi K[3], Komaki H[4], Taniguchi-Ikeda M[5], Ishigaki K[6], Toda T[1]

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205P Early diagnosis for Duchenne muscular dystrophy (DMD) to shape and influence the model of care to support children and families following an early diagnosis of DMD

Lee S[1], Bradley C[1], Evans K[1], Lorentzos M[2], Servais L[3], Parsons J[4]

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206P A quality improvement proposal: improving care management of complex neuromuscular patients – establishing a neuromuscular complex care centre as part of the translational neuromuscular pathway

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207P Mobile muscle clinics operating from a UK centre of excellence: report of patient experience and quality of care

Kulshrestha R[1], Emery N[1], Strachan K[1], Willis T[1], Bassie C[1]

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208P Are people living with neuromuscular disorders in the north of England satisfied with National Health Service wheelchair service provision?

Michell-Sodhi J[1], Moat D[1], Dias T[1], Mason J[1], Robinson E[1], Schiava M[1], Barr E[1], Bolano C[1], Doaa S[1], Wong K[1], Maha E[1], Emma G[1], Michela G[1], Michelle M[1], Tasca G[1], Diaz-Manera J[1], James M[1], Straub V[1], Marini-Bettolo C[1], Muni-Lofra R[1]

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209P Custom orthosis improves mobility and caregiver experience in an adolescent with Congenital Myasthenic syndrome and Myofibrillar Myopathy

Varghese V[1], Rajadurai I[1], Paul M[1], Banavara Shyamprasad S[2], Mathew A A[1,2]

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210P Advancing upper limb motor function evaluation in Duchenne muscular dystrophy and Spinal Muscular Atrophy via kinematic parameterization with the wearable device "ArmTracker"

Natera De Benito D[1], Favata A[2], Expósito-Escudero J[1], Gallart R[3], Moya O[1], Roca S[1], Marzabal Gatell A[2], van Noort L[2], Ortez C[1], Torras C[3], Nascimento A[1], Medina-Cantillo J[1], Pàmies Vilà R[2], Font-Llagunes J[2]

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211P Following patient mobility in daily life: the EJP-DT4RD project

Hogrel J[1], Muni-Lofra R[2], Santmarty P[1], Decostre V[1], Marques T[1], Haf Davies E[3], Straub V[2], the DT4RD Project Group

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212P Identifying biomarkers for prediction of fall-risk in patients with Inclusion Body Myositis (IBM)

Hunn S[1], Weihl C[1]

[1]Washington University School of Medicine

213P Sensitivity to change of the Motor Function Measure (MFM) in myotonic dystrophy type 1

Ribault S[1], Rippert P[1], Bassez G[2], Duong T[3], Vuilllerot C[1]

[1]Hospices Civils De Lyon, [2]Stanford University, 3Hôpital de la Pitié Salpêtrière

214P The 10-meter model: predicting the 6-minute walk test in Pompe disease

El Kaim A[1], Fer F1, Hogrel J1

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215P Evaluating the responsiveness of patient reported outcome measures (PROs) to change in valosin-containing protein multisystem proteinopathy (MSP1) over 24 months

Iammarino M[1], Reash N[1], Lowes L[1,2], Pietruszewski L[3], Adderley K[1], Humphrey L[1], Beale A[1], Steiner C[1], Smith M[1], Alfano L[1,2]

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216P A questionnaire-based investigation into levels physical disability and of physical activity in adults with neuromuscular disease in a UK neuromuscular centre

Willis T[1,2], *Strachan K*[1], *Emery N*[1]

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217P Comparing IBMFRS and sIFA as progression indicators in Inclusion Body Myositis patients from the INSPIRE-IBM trial

Gaid P[1], *Wencel M*[1], *Hernandez I*[1], *Goyal N*[1], *Dimachkie M*[2], *Lloyd T*[3], *Mohassel P*[3], *Weihl C*[4], *Freimer M*[5], *Shaibani A*[6], *Wicklund M*[7], *Dixon S*[8], *Chahin N*[9], *Wang L*[10], *Shieh P*[11], *Amato A*[12], *Quinn C*[13], *Carbunar O*[14], *Mozaffar T*[1], *INSPIRE-IBM Study Group*[1]

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218P Inter-rater reliability of adapted test of neuromuscular disorders (ATEND) wheelchair-based assessment

Nelson L[1], *Tang W*[2], *Pasternak A*[3], *Glanzman A*[4], *Muni Lofra R*[5], *Duong T*[2]

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219P Enhancing clinical trial eligibility criteria in FSHD: validating whole-body MRI as a key outcome measure

Widholm P[1,2,3], *Karlsson M*[1], *Pini J*[4,5], *Puma A*[4], *Villa L*[4], *Cavali M*[4], *Ezaru A*[4], *Bassez G*[6], *Marty B*[6], *Evangelista T*[6], *Thomas R*[7,8], *Danjoux L*[7,8], *Tard C*[7,8], *Sacconi S*[4,5]

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220VP Wearable sensors to evaluate and monitor neuromuscular patients in real world environment

Diella E[1], *Storm F*[1], *Molteni L*[1], *Delle Fave M*[1], *Canella G*[1], *Meola G*[2], *Biffi E*[1], *D'Angelo M*[1]

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221P Initial data from the achieve trial of DYNE-101 in adults with myotonic dystrophy type 1 (DM1)

Wolf D[1], *Lilleker J*[2], *Bassez G*[3], *Diaz-Manera J*[4], ***Kools J***[5], *Pane M*[6], *Roxburgh R*[7], *Schoser B*[8], *Turner C*[9], *Mix C*[1], *Ray S*[1], *Han B*[1], *Farwell W*[1], *Sansone V*[10]

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222P Development of a standardized information model for rare neuromuscular diseases

Gazzerro E[1], *Hübner M*[2], *Nyoungui E*[3], *Krefting D*[3], *Spuler S*[1], *Zschüntzsch J*[3], *Schepers J*[2], *Röttger R*[4]

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223P Mental health support for children and young people with Duchenne muscular dystrophy – who, when and how across the UK

Geagan C[1], *Sandhu A*[1], *Bouquillon L*[2], *Conn R*[3], *Bindman D*[2], *Pattni J*[2], *Turner C*[1], *McDonald R*[2], *Alex J*[5], *Rodney S*[4], *Quinlivan R*[2], *Guglieri M*[1], *Straub V*[1]

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224P Psychosocial discussions in neuromuscular clinics from a professional lens: evidence from a service evaluation regarding barriers to discussions

Geagan C[1], *Sandhu A*[1], *Mason J*[1], *McCallum M*[1], *Muni-Lofra R*[1], *Moat D*[1], *Wong K*[1], *Robinson E*[1], *Grover E*[1], *Michel-Sodhi J*[1], *Bolaño Diaz C*[1], *Schiava M*[1], *Salman D*[1], *James M*[1], *Tasca G*[1], *Diaz Manera J*[1], *Guglieri M*[1], *Straub V*[1], *Elseed M*[1], *Marini Bettolo C*[1]

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225P Initial data from the DELIVER trial of DYNE-251 in males with DMD Mutations amenable to Exon 51 skipping

De Waele L[1], *Campbell C*[2], *Deconinck N*[3], *Flanigan K*[4], *Lorentzos M*[5], *Phan H*[6], *Shieh P*[7], *Mix C*[8], *Ray S*[8], *Wang D*[8], *Farwell W*[8], *Dugar A*[8], *Naylor M*[8], *Guglieri M*[9], *on behalf of the DELIVER study investigators*

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226P Phase 3, randomized, global study assessing efficacy and safety of del-desiran for the treatment of Myotonic Dystrophy Type 1: HARBOR trial design

Fowler M[5], *Johnson N*[1], *Thornton C*[2], *Day J*[3], *Sansone V*[4], *McEvoy B*[5], *Tai L*[5], *Knisely B*[5], *Brandt T*[5], *Gallagher K*[5], *Hughes S*[5], *Ackermann E*[5]

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227P Advancing neuromuscular education and care within physiotherapy: POD-NMD (Physiotherapy Online Delivery - Neuromuscular Diseases)

Slim C[1], *Mayhew A*[2]

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228P Don't lose your HEAD: a quality improvement proposal for early recognition of neuromuscular diseases in floppy neonates in the North of England

Reeves T[1,2], *Muni-Lofra R*[1], *Harris L*[1], *Richardson R*[2], *Guglieri M*[1], *Marini-Bettolo C*[1]

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229P Home mechanical ventilation in paediatric neuromuscular disorders in a resource limited setting

Ramesh Babu R[1,2], *Kinimi I*[3], *Shinde S*[3], *Mohan Rao N*[3], *Sahoo A*[3], *Maganthi M*[1], *Agnes Mathew A*[1,2]

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230P Physiotherapy for rare hereditary NMDs- an open and free e-learning course for physiotherapists. Results after 18 months online at sjelden.no

Rosenberger A[1], *Schandy R*[2], *Lysne S*[2], *Hæstad H*[1]

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231P Efficacy of tranilast in preventing exacerbating cardiac function and death from heart failure in Muscular dystrophy patients with advanced-stage heart failure: a single-arm, open-label, multicenter study

Matsumura T[1], *Fukudome T*[2], *Motoyoshi Y*[3], *Nakamura A*[4], *Kuru S*[5], *Segawa K*[6], *Kitao R*[7], *Watanabe C*[8], *Tamura T*[9], *Takahashi T*[10], *Hashimoto H*[11], *Sekimizu M*[11], *Saito A*[11], *Asakura M*[12], *Kimura K*[13], *Iwata Y*[14]

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232VP Patient knowledge of the risks of glucocorticoid management in a specialist adult muscle clinic

Kent L[1], *Begeti F*[1], *Turner H*[1], *Brady S*[1]

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233VP Review of inpatient care of children affected by neuromuscular disorders or complex neurodisability from the carer perspective

Thomas S[1,2], *Gallagher S*[1,2], *Khalid A*[1,2], *Alhaswani Z*[1,2]

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267P-279P: Cell insights, muscle homeostasis

267P Circulating alpha-Klotho combats TGF-beta-induced sarcopenia

Ohsawsa Y[1], *Nishimatsu S*[1], *Saito F*[2], *Sunada Y*[1]
[1]*Kawasaki Medical School,* [2]*School of Medicine, Teikyo University*

268P Age and sex affect human skeletal muscle secretome

Crisol B[1], *Pinzon N*[1], *Orio J*[1], *Gaut-Serey L*[1], *Ohana J*[1], *Bensalah M*[1], *Peterson C*[2], *Butler-Browne G*[1], *Mouly V*[1], *Bigot A*[1], *Trollet C*[1]
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269P Human neuroblastoma cell-derived neurons and iPSC-derived neurons as models for neuromuscular disorders

Wang H[1], *Pommerenke C*[1], *Hauer V*[1], *Rand U*[1], *Eberth S*[1], *Nagel S*[1], *Dirks W*[1], *Werr L*[2,3], *Fischer M*[2,3], *Steenpaß L*[1]
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270P Single nucleus RNA sequencing reveals unique myonuclei populations in Late-Onset Myopathy

Soule T[1], *Pontifex C*[1], *Rosin N*[1,2], *Joel M*[1], *Lee S*[1], *Nguyen M*[1,3], *Chhibber S*[3], *Pfeffer G*[1,3,4]
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271P Exploring the effects of the epi-drug Remodelin on murine myoblasts differentiation

Sian V[1,2], *Sarparanta J*[1,3], *Hentschel A*[4], *Jonson P*[1,3], *Roos A*[5,6], *Natraj Gayathri S*[1,3], *Udd B*[1,7], *Savarese M*[1,3], *Nebbioso A*[2]
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272P High phosphate levels induce atrophy and FGF23 expression in skeletal muscle

Heitman K[1], *Fajol A*[1], *Thomas M*[1], *Li Q*[1], *Campos I*[1], *Westbrook D*[1], *Komarova S*[1], *Alexander M*[1], *White K*[2]
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273P Estrogen deficiency results in reduced maximal running capacity without changes in the phosphorylation of myosin regulatory light chain in female rats

Karvinen S[1], *Lee E*[1,2], *Nissinen T*[1], *Ylä-Outinen L*[1], *Jalkanen A*[3], *Karppinen J*[1,4], *Vieira-Potter V*[5], *Lipponen A*[1]
[1]*University of Jyväskylä,* [2]*University of Montréal,* [3]*University of Eastern Finland,* [4]*University of Helsinki,* [5]*University of Missouri*

274P Generation of cardiac organoids from DuchenneMuscularDystrophy patient-derived induced pluripotent stem cells: a novel approach to understanding cardiomyopathy

Przymuszała M[1], *Stępniewski J*[1], *Florczyk-Soluch U*[1], *Dulak J*[1]
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275P Feasibility of assessing muscle oxygenation in patients with neuromuscular disease using near-infrared spectroscopy

Tang W[1], *Salvatore S*[1], *Khonde S*[1], *Montalvo S*[1], *de Monts C*[1], *Smith S*[1], *Hageman N*[1], *Blumberg Y*[1], *Ataide P*[1], *Ni Ghiollagain N*[1], *Parker D*[1], *Wheeler M*[1], *Day J*[1], *Christle J*[1], *Duong T*[1]
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276P Exploring muscle endurance in a neuromuscular population: insights from the assisted 6-minute cycling test combined with cardiopulmonary exercise testing and near-infrared spectroscopy

Tang W[1], *de Monts C*[1], *Montalvo S*[1], *Dunaway Young S*[1], *Salvatore S*[1], *Khonde S*[1], *Smith S*[1], *Hageman N*[1], *Blumberg Y*[1], *Ataide P*[1], *Ni Ghiollagain N*[1], *Parker D*[1], *Wheeler M*[1], *Day J*[1], *Wheeler M*[1], *Duong T*[1]
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277P Single-nuclei RNAseq in myotonic dystrophy type I: DMPK accumulation and myofiber repair

Todorow V[1], *Hayashi S*[2], *Meinke P*[1], *Schoser B*[1], *Nishino I*[2]
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278P Mapping human skeletal muscle enhancers to increase rates of genetic diagnosis

Taylor R[1,2], *Taylor J*[1,2], *Denisenko E*[1,2], *Jones M*[1,2], *Clayton J*[1,2], *Laing N*[1,2], *Forrest A*[1,2], *Alinejad-Rokny H*[3], *Ravenscroft G*[1,2]

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279P Exploring myogenic tremor in an animal model of MYBPC1-Associated Myopathy: a comprehensive study

Inashkina I[1], *Zdanovica A*[1], *Lunge M*[1], *Zayakin P*[1], *Upite J*[2], *Dzirkale Z*[2], *Alnis J*[4], *Lace B*[3], *Jansone B*[2], *Stavusis J*[1]

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361P-400P: DMD - imaging and outcome measures

361P Development of a next-generation multiplex ddPCR assay for measurement of in-frame dystrophin mRNA in people with DMD treated with BMN 351

Tamura S[1], *Sidhu I*[1], *Ilagan J*[1], *Yu A*[1], *Byer A*[1], *Kamath S*[1], *Staskus L*[1], *Russell C*[1], *Melton A*[1], ***Larimore K***[1]

[1]*BioMarin Pharmaceutical Inc*

362P Whole-body MRI reveals common and distinctive muscle involvement in "clinically asymptomatic" female carriers of pathogenic DMD variants

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363P A cross-sectional survey upper limb functional using PUL 2.0 at large clinical centers and registries in patients with Duchenne muscular dystrophy

Coratti G[1], *Niks E*[2], *van der Holst M*[3], *Tian C*[4], *Mercuri E*[1], *Muntoni F*[5,6], *Servais L*[7,8], *Ward S*[9]

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364P Gross motor delays in boys with Duchenne muscular dystrophy are seen in infancy

Lowes L[1,2], *Reash N*[1], *Iammarino M*[1], *Connolly A*[2], *Pietruszewski L*[3], *Smith M*[1], *Peng J*[4], *Steiner C*[1], *Tsao C*[2], *Waldrop M*[1,2], *Flanigan K*[1,2], *Chagat S*[1], *Meyer A*[1], *Mendell J*[5], *Alfano L*[1,2]

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365P Duchenne video assessment 2.0 scorecard performance: evaluation of inter-item and inter-scorecard relationships

Contesse M[1], *Sapp A*[1], *Zigler C*[2], *Chen W*[1], *Marshall J*[1], *Gensler G*[1], *Brown C*[1], *Barnes R*[1], *King D*[1], *Wilson S*[1], ***Leffler M***[1]

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366P Social Vulnerability and Fluid Cognition in Duchenne Muscular Dystrophy: The Impact on Boys and Carrier Mothers

Penumalee V[1], *Karra H*[2], *Javalkar S*[1], *Arun S*[1], *Kaat A*[3], *Thangarajh M*[1]

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367P Mortality among males with muscular dystrophy in the USA: estimation from whole-population cause-of-death records

Hayes E[1], *Giafaglione J*[1], *Signorovitch J*[2], *Sajeev G*[2], *Zhu A*[2], *Shell R*[1], *Waldrop M*[3], *Cripe L*[1], *Wright L*[1], *Nandi D*[1]

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368P Ankle contractures in Duchenne muscular dystrophy: using real world data to study the influence of the gastrocnemius and soleus muscles on functional decline

Fleerakkers E[1,2,3], *Pelsma M*[4], *Krom Y*[1,3], *Hoek R*[1,3], *Kan H*[5], *van Zwet E*[6], *Houwen-van Opstal S*[3,4], *Niks E*[1,3], *van der Holst M*[2,3]

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369P Elbow function and thumb pinch strength are relevant for activities of daily living for non-ambulant DMD patients

Michaëls M[1,2], *Naarding K*[1,2], *van der Holst M*[1,2], *van Zwet E*[1], *Kan H*[2,3], *Niks E*[1,2]

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370P Gait analysis in children with Duchenne muscular dystrophy: overground vs. treadmill walking

Deconinck N[1], *Ruiz P*[2], *Kaleeta J*[3], *Cheron G*[2], *Cebolla A*[2]

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371P Screening for psychosocial needs in adults with Duchenne muscular dystrophy: preliminary findings on using the GAD7 and PHQ9

Bouquillon L[1], *Pattni J*[1], *Bindman D*[1], *Geagan C*[2], *Conn R*[3], *McDonald R*[1], *Turner C*[2], *Rodney S*[4], *Johnson A*[5], *Guglieri M*[2], *Straub V*[2], *Quinlivan R*[1]

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372VP MRI fat fraction in Duchene muscular dystrophy (DMD): inter-reader variability of the 90th percentile fat fraction value

Hammond M[1], *Harris J*[1], *Mesbah S*[1], *Roche F*[2], *Berger M*[1], *Zabbatino S*[1], *Scheyer R*[3], *Holland S*[1]

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373P Association between the time of diagnosis and muscle breakdown in DMD patients diagnosed under two years of age

Coşkun A[1], *Topaloğlu H*

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374P Relationship between the Pediatric Outcomes Data Collection Instrument (PODCI) and EQ-5D-3L utility score in Duchenne muscular dystrophy

Posner N[1], *Merla V*[1], *Aslam Z*[1], *Bushmakin A*[1], *Cappelleri J*[1]

[1]*Pfizer, Inc.*

375P Relationship between Pediatric Outcomes Data Collection Instrument (PODCI) and North Star Ambulatory Assessment (NSAA)

Posner N[1], *Merla V*[1], *Aslam Z*[1], *Bushmakin A*[1], *Cappelleri J*[1]

[1]*Pfizer, Inc.*

376P Ataluren in patients with nonsense mutation DMD (nmDMD): retrospective evaluation from a tertiary referral center in Turkey

Öz Yıldız S[1,3], *Bulut N*[2], *Gürbüz İ*[2], *Öztoprak Ü*[1], *Tunca Yılmaz Ö*[2], *Haliloğlu G*[1]

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377P Patterns of disease progression in Duchenne muscular dystrophy: heterogeneous changes in motor function tasks among patients with similar trajectories over one year

Posner N[1], *Talaga A*[1],*Johnson M*[2], *Gomez-Lievano A*[2], *Akbarnejad H*[2], *Ma Y*[2], *Aslam Z*[1], *Kane A*[1], *Bhambri R*[1], *Dukacz S*[3], *Cappelleri J*[1], *Signorovitch J*[2]

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378P Geospatial driver of cognition in biological mothers of sons with Dystrohinopathy

Javalkar S[1], *Penumalee V*[1], *Arun S*[1], *Karra H*[1,2], *Kaat A*[3], *Thangarajh M*[1]

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379P A qualitative study on people with Duchenne muscular dystrophy and caregivers' experiences during the transition process from pediatric to adult healthcare

Marcassoli A[1], Moroni I[2], Guastafierro E[1], Brigliadori B[2], Nardocci N[2], Leonardi M[1], De Angelis F[3], Langer T[4], Rodger S[4], Willems J[4], Kraus De Camargo O[5], Frei J[5], Swain A[5], Ringer D[5], Gorter J[5], Pozniak K[5], Rajapakse N[5], Fournier A[6], Gutierrez R[6], Osman H[7], Reeskau G[8], McCauley D[5], Wang E[5], Friedrich S[4]

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380VP Assessment of lung function and respiratory muscles using ultrasonography in Duchenne muscular dystrophy

Ferreira Lima K[1], Arruda P[1], Albuquerque M[1], Zanoteli E[1]

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381P Survey results for use of the Performance of the Upper Limb Module in patients with Duchenne muscular dystrophy: UK North Star national registry

Wolfe A[1,2], Brooke M[2], Aslam N[2], Manzur A[1,2], Muntoni F[1,2], Baranello G[1,2]

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382P Decreased quality of life in Duchenne muscular disease patients related to functional neurological and cardiac impairment

Jurikova L[1], Masarova L[2,11], Panovsky R[2,3], Pesl M[2,3,4], Zondra Revendova K[5,6], Volny O[2,5,6], Feithova V[2,7], Holecek T[2,7,8], Kincl V[2,3], Danhofer P[1], Vohanka S[9], Haberlova J[10], Podolska K[10]

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383P A qualitative evaluation of meaningful change on the north star ambulatory assessment and performance of upper limb in Duchenne muscular dystrophy

Murphy A[1], Ciobanu T[2], Davies E[2], Gillman A[3], Barrett L[3], Johnson A[4], Mills J[3], Heinrich P[3], Przydzial K[3], Ewens B[3], Vandenberg G[3], Cano S[3], Mayhew A[5]

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384P Functional Outcome Measures (FOMs) guiding clinical care of children with Duchenne muscular dystrophy

Khandekar G[1], Banavara Shyamprasad S[1], Ramesh Babu R[1,2], Kumar A[2], Satyam P[1,2], Maganthi M[2], Krishna G[1], Mathew A[1,2]

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385P Quantitative magnetic resonance of masticatory muscles in adolescents with Duchenne muscle dystrophy

Avila Smirnow D[1,2,3], Meza C[1,2], González C[1,2], Uribe S[4,5], Salinas L[1,2]

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386P Dark-adaptation visual thresholds in Duchenne muscular dystrophy patients with genetic backgrounds affecting different dystrophin proteins

Dias S[1], Barboni M[1,2], Brasil A[1], Lima K[3], Camelo C[3], Sindeaux R[3], Resende M[3], Albuquerque M[3], Zanoteli E[3], Ventura D[1]

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387P Quantifying function from birth through adulthood in Duchenne muscular dystrophy: a cross-sectional evaluation of the Neuromuscular Gross Motor Outcome

Iammarino M[1], Reash N[1], Smith M[1], Steiner C[1], Beale A[1], Humphrey L[1], Lowes L[1], Alfano L[1]

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388P Remote evaluation of functional outcomes in Duchenne muscular dystrophy: navigating opportunities and obstacles

Banavara Shyamprasad S[1], *Khandekar G*[1], *Ramesh Babu R*[1,2], *Krishna G*[1], *Mathew A*[1,2]
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389P Natural history of bone health in Duchenne muscular dystrophy: A systematic review and implications for the design of a clinical trial

De Ford C[1], *Guridi M*[1], *Chen Y*[2], *Murphy A*[3], *Wood C*[4], *McMillan H*[5], *Mercuri E*[6], *Crabtree N*[7], *Ward L*[8,9]
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390P Assessing biomarkers of bone metabolism and the role of the IL-6 signalling pathway in patients with Duchenne muscular dystrophy

De Ford C[1], *Guridi M*[1], *Fruechtenicht C*[1], *See C*[1], *Houghton R*[1], *Chen Y*[2], *Murphy A*[3], *Wood C*[4], *Ward L*[5,6], *Crabtree N*[7], *Mercuri E*[8], *McMillan H*[9]
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391P Agreement and accuracy of ambulatory definitions in Duchenne muscular dystrophy (DMD): a cross-sectional analysis

Macedo A[1], *Logan J*[1], *Dai D*[1], *Ricchetti-Masterson K*[1]
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392P Investigating the data landscape of Duchenne muscular dystrophy: Answering your research questions with the right data source

Furby H[1], *Johnson M*[1], *Signorovitch J*[2], *Moride Y*[3], *Castillon G*[3], *Simpson A*[4]
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393P Understanding oropharyngeal dysphagia in Duchenne muscular dystrophy: A 10-year longitudinal cohort study

Scholten S[1], *Lagarde M*[1], *Knuijt S*[2], *Houwen-van Opstal S*[1], *Erasmus C*[3], *Groothuis J*[2]
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394P Longitudinal changes in the North Star Ambulatory Assessment and leg muscle fat fraction in DMD

Forbes S[1], *Nair K*[2], *Willcocks R*[1], *Barnard A*[1], *Lott D*[1], *Senesac C*[1], *Rock K*[1], *Finanger E*[3], *Brandsema J*[4], *Subramony S*[1], *Wang D*[4], *Rooney W*[5], *Walter G*[1], *Vandenborne K*[1]
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395P Age at loss of ambulation in patients with nmDMD from the STRIDE Registry: sensitivity analyses

Muntoni F[1], *Buccella F*[2], *Nascimento Osorio A*[3], *Tulinius M*[4], *Bernadete Dutra de Resende M*[5], *Johnson S*[6], *Werner C*[7], *Anbu B*[6], *Liu E*[6], *Mercuri E*[8]
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396P 6-year follow-up on muscle affection in female carriers of Dystrophinopathy

Lyu Z[1], *Poulsen N*[1], *Joensen H*[1], *Dunø M*[2], *Bundgaad H*[3], *Vejlstrup N*[3], *Lando C*[1], *Hansen A*[4], *Vissing J*[1]
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397P 6-year follow-up on cardiac affection in female carriers of Dystrophinopathy

Lyu Z[1], *Poulsen N*[1], *Joensen H*[1], *Bundgaad H*[2], *Vejlstrup N*[2], *Lando C*[1], *Hansen A*[3], *Vissing J*[1]
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398P Magnetic resonance imaging detects bone alterations in corticosteroid treated boys with DMD

Kunnath Ravindranunni R[1], *Grewal R*[2], *Cottrell C*[2], *Bernier A*[1], *Tuna I*[1], *Vandenborne K*[1], *Walter G*[1], *Rajapakse C*[2], *Willcocks R*[1]
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399P MRI assessment of microdystrophin gene therapy in DMD: a five year longitudinal study

Willcocks R[1], *Lott D*[1], *Forbes S*[1], *Vandenborne K*[1], *Walter G*[1]
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400P Longitudinal evaluation of ambulatory function with ankle wearable technology in ambulant DMD

Poleur M[1], *Parinello G*[2], *Vrščaj E*[3], *Kumhera M*[4], *Bisson C*[2], *Aragon-Gawińska K*[5], *Anghelescu C*[6], *Daron A*[1], *Szabo L*[7], *Leanca M*[8,9,10], *Mirea A*[8,9,10], *Kodsy S*[11], *Saleh A*[11], *Osredkar D*[3], *Haberlova J*[4], *Potulska-Chromik A*[5], *Butoianu N*[6,8], *Eggenspieler D*[2], *Strijbos P*[12], *Servais L*[13,14]
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538P-577P, 578VP: Acquired, inflammatory, myositis

538P Muscle haptoglobin biomarks cachexia induced by anti-acute myeloid leukemia chemotherapy

Rybalka E[1,2,3], *Campelj D*[1,4], *Spiesberger G*[1,2], *Formosa L*[5], *Steele J*[6], *Zhang H*[6], *Schittenhelm R*[6], *Leow L*[3], *Bhandari N*[1], *Goodman C*[7], *Timpani C*[1,2,3]
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539P A late-onset multiple-acyl-CoA-dehydrogenase deficiency-like condition is more common in West of Scotland than primary genetic MADD, and is likely multifactorial

Watson-Fargie T[1], *Coomber A*[2], *Barr M*[3], *Brennan K*[1], *Dale J*[4], *Fletcher E*[5], *Gallagher P*[1], *Molinari E*[1], *Miller-Hodges E*[6], *O'Sullivan D*[7], *Stewart K*[8], *Taylor R*[9], *Topf A*[10], *Straub V*[10], *Edwards R*[3], *Stewart W*[11], *Farrugia M*[1], *Longman C*[8]
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540P Assessing the effect of chronic nitrate supplementation on muscle mass and physical function outcomes

Edwards H[1,2], *Morton J*[2], *Moseley J*[3], *Marshall T*[3], *Berry A*[2], *Igwesi-Chidobe C*[3], *El-Khamisy S*[2], *Jones H*[2], *Farrow M*[4]
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541P Comparative evaluation of pulmonary function tests and self-reported respiratory function in Inclusion Body Myositis based on NT5c1A antibody serology status

Wencel M[1], *Bjazevic K*[1], *Goyal N*[1], *Carbunar O*[2], *Freimer M*[14], *Dimachkie M*[3], *Quinn C*[4], *LLoyd T*[5], *Mohassel P*[5], *Weihl C*[6], *Shaibani A*[7], *Wang L*[8], *Chahin N*[9], *Amato A*[10], *Wicklund M*[11], *Shieh P*[12], *Herbelin L*[13], *Barohn R*[13], *Mozaffar T*[1]

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542P INSPIRE-IBM: an NIH-funded, two-year, multicenter, observational study in inclusion body myositis (IBM)-an update

Wencel M[1], *Goyal N*[1], *Carbunar O*[2], *Freimer M*[3], *Dimachkie M*[4], *Quinn C*[5], *Lloyd T*[6], *Mohassel P*[6], *Weihl C*[7], *Shaibani A*[8], *Wang L*[9], *Nizar C*[10], *Amato A*[11], *Wicklund M*[12], *Dixon S*[12], *Shieh P*[13], *Herbelin L*[14], *Herbelin L*[14], *Mozaffar T*[1]

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543P Riboflavin-responsive MADD, a modern day problem?

Hustinx M[1], *Brady S*[2]

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544P A single center Indian cohort of Inclusion Body Myositis

George J[1], *Rajalakshmi P*[2], *Sundaram S*[1], *Nair S*[1]

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545P Capillary abnormalities in Immune-Mediated Necrotizing Myopathy: more than collateral damage?

Eisenlohr P[1,2], *Preusse C*[1], *Roos A*[4], *Stenzel W*[1], *Kleefeld F*[2]

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546VP Deep immunoprofiling in inclusion body myositis and trajectory analysis

Roy B[1], *DiStasio M*[1], *Hackbarth R*[2], *Bahrassa F*[1], *Joo D*[1], *Pham M*[1], *O'Connor K*[1]

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547P The diagnostic value of serum periostin for myocardial involvement in immune-mediated necrotizing myopathy

Xu L[1], *Yang M*[1], *Kosari M*[2], *Li Y*[1], *Bu B*[1], *Ji S*[1]

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548P A role of heparin-binding epidermal growth factor-like growth factor in myocardial involvement in immune-mediated necrotizing myopathy

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549VP High dose intravenous corticosteroids in refractory Immune Checkpoint Inhibitor-related Myocarditis/Myositis/Myasthenia overlap syndrome

Forcina F[1], *Tufano L*[1], *Lauletta A*[1], *Morino S*[1], *Musumeci B*[2], *Antonini G*[1], *Garibaldi M*[1]

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550P Immune checkpoint inhibitor-induced myositis in a patient with cholangiocarcinoma

Joe S[1], ***Kim S***[1], *Choi Y*[1], *Choi Y*[1], *Park H*[1]

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551P BiP (GRP78) as potential blood and tissue biomarker in systemic sclerosis

Preusse C[1,2,3], Dobelmann V[4], Kleefeld F[1], Hentschel A[5], Holla E[6], Ruck T[4], Stenzel W[3], Siegert E[7], Roos A[4,6,8]
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552P Binding immunoglobulin protein (BiP) abundance and distribution in Idiopathic Inflammatory Myopathies (IIM)

Kleefeld F[1], Preusse C[1], Hentschel A[4], Stenzel W[1], Dobelmann V[2], Roos A[2,3,5]
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553P Brachio-cervical Inflammatory Myopathy, an unknown disease with diverse serological and histopathological findings and poor response to treatment

Kapetanovic Garcia S[1,2], Jimenez-Almonacid J[3], Toldos-Gonzalez O[3], Ruiz-Lucea E[4], Rodrigo-Armenteros P[1,2], Hernandez-Lain A[3], Dominguez-Gonzalez C[5,6]
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554P Mitochondrial pathology in Myositis: a multicentric case series

Lauletta A[1], Bosco L[2], Merlonghi G[1], Falzone Y[2], Cheli M[3], Bencivenga R[4], Léonard-Louis S[5], Benveniste O[6], Ruggiero L[4], Maggi L[3], Previtali S[2], Garibaldi M[1]
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555P Assessment of IBM-FRS total score and specific domains in a large cohort of Inclusion Body Myositis patients

Vicino A[1,2], Lauletta A[3], Barbaccia A[4], Valentino L[5], Cheli M[6], Saccani E[7], Grandis M[8], Coccia M[9], Barp A[10], Ravaglia S[11], Bortolani S[12], Ruggiero L[13], Mongini T[14], Verriello L[15], Vattemi G[6], Filosto M[16], Liguori R[5], Rodolico C[4], Garibaldi M[3], Maggi L[1]
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556P Contractile properties of permeabilised single muscle fibres from female patients with Idiopathic Inflammatory Myopathies

Henning F[1], Kohn T[2]
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557P Skin manifestations of immune-mediated necrotizing myopathy with anti-HMGCR antibody

Kurashige T[1], *Nakamura R*[2], *Murao T*[1], *Mine N*[4], *Sato M*[3], *Katsumata R*[1], *Kanaya Y*[1], *Dodo Y*[1], *Sugiura T*[1], *Ohshita T*[1]
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558P Contactin-1 antibody-associated chronic inflammatory demyelinating polyneuropathy (CIDP) in a pediatric patient: beyond the tip of an iceberg

Özel E[1], *Öncel İ*[1], *Öztoprak Ü*[1], *Temuçin Ç*[2], *Haliloğlu G*[1]
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559P Immunological biomarkers in immune-mediated and hereditary polyneuropathies

Kodal L[1], *Krag T*[1], *Dysgaard T*[1]
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560VP Immune-mediated necrotizing myopathy in 14 Polish patients – clinical course and treatment outcome

Rajczewska Oleszkiewicz C[1], *Kierdaszuk B*[1], *Kostera-Pruszczyk A*[1]
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561P PDGFRa as a reliable surface marker for fibro-adipogenic progenitor (FAP) cells

Park J[1], *Walli S*[1], *Bachir D*[1], *Dobelmann V*[1], *Preuße C*[2], *Stenzel W*[2], *Brunn A*[3], *Meuth S*[1], *Ruck T*[1], *Nelke C*[1]
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562P Rare mimics of idiopathic inflammatory myopathies – a single centre cohort from India

Sivaraman Nair S[1], *George J*[1], *Poyuran R*[1]
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563P Abatacept, the solution for a patient with immune-mediated necrotizing myopathy

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564VP Investigating motor and bulbar severity in NT5c1A seropositive and seronegative IBM participants in the INSPIRE-IBM trial

Herrera M[1], *Wencel M*[1], *Hernandez I*[1], *Goyal N*[1], *Dimachkie M*[2], *Lloyd T*[3], *Mohassel P*[3], *Weihl C*[4], *Freimer M*[5], *Shaibani A*[6], *Wicklund M*[7], *Dixon S*[8], *Chahin N*[9], *Wang L*[10], *Shieh P*[11], *Amato A*[12], *Quinn C*[13], *Carbunar O*[14], *Mozaffar T*[1], *INSPIRE-IBM Study Group*[1]
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565VP Investigating highly differentiated cytotoxic T cells and functional severity in participants with Inclusion Body Myositis in the INSPIRE-IBM trial

Herrera M[1], *Wencel M*[1], *Hernandez I*[1], *Goyal N*[1], *Dimachkie M*[2], *Lloyd T*[3], *Mohassel P*[3], *Weihl C*[4], *Freimer M*[5], *Shaibani A*[6], *Wicklund M*[7], *Dixon S*[8], *Chahin N*[9], *Wang L*[10], *Shieh P*[11], *Amato A*[12], *Quinn C*[13], *Carbunar O*[14], *Mozaffar T*[1], *INSPIRE-IBM Study Group*[1]
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566P CIC-1 chloride channel inhibition improves disease symptoms and survival in a rat model of muscle-specific kinase (MuSK) Myasthenia Gravis

Skals M[1], *Morgen J*[1], *Huus N*[1], *Skov M*[1], *Kelly N*[1], ***Broch-Lips M***[1]
[1]*NMD Pharma A/S*

567P A prospective observational study on the effect of intravenous immunoglobulin on small fiber neuropathy after SARS-CoV-2 infection or vaccination

Shin J[1], *Lee J*[2]
[1]*Hallym Medical Center, Dongtan Sacred Heart University,* [2]*Kyungpook National University Hospital*

568P Unraveling interferon-related autophagic signatures in Immune Myopathies

Authier F[1], *Hou C*[1], *Periou B*[1], *Souvannanorath S*[1], *Malfatti E*[1], *Martin L*[1]
[1]*Paris Est-creteil University*

569P Revisiting Systemic Sclerosis Myopathy: unveiling unique histopathological signatures and overlaps with immune myopathies features

Authier F[1], *Zaidan L*[1], *Le Gouellec N*[2], *Periou B*[3], *Relaix F*[1], *Mouthon L*[4], *Hachulla E*[5]
[1]*Paris Est-creteil University,* [2]*Hospital of Valenciennes,* [3]*Henri Mondor University Hospital, APHP,* [4]*Cochin University Hospital, APHP,* [5]*Lille University Hospital*

570P Physiological and molecular mechanisms underlying chronic alcoholic-related myopathy

Baumann C[1], *Ganjayi M*[1], *Brown A*[1], *Willis C*[2]
[1]*Ohio University,* [2]*University of Bradford*

571P Muscular metabolic plasticity in 3D in vitro models against systemic stress factors in ME/CFS and long COVID-19

Mughal S[1,4], *Andújar-Sánchez F*[3,4], *Sabater-Arcis M*[1], *Fernández-Costa J*[1], *Ramón-Azcón J*[1,2]
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572P Dysphagia pattern in Inclusion Body Myositis as a distinguishing feature: insight from a patient initially presenting with rapidly progressing dysphagia

Park Y[1,2], *Bae J*[3,4], *Jung Y*[3,4]
[1]*Department of Neurology, Pusan National University Hospital,* [2]*Department of Neurology Pusan National University School of Medicine,* [3]*Department of Neurology Kangdong Sacred Heart Hospital,* [4]*Department of Neurology Hallym University College of Medicine*

573P The interlocution of autophagy and interleukin-6 in immune-mediated necrotizing myopathy

Liang W[1,2,3], *Chang S*[1], *Lin P*[5], *Jong Y*[1,3,4], *Liu H*[5]
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574P Infectious muscles diseases: a comprehensive case study of 7 patients from Indian cohort

Sharma M[1], *Bhatia R*[1], *Venugopalan Y V*[1]
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575P FORTIFY: a phase 3 study to evaluate efficacy & safety of BBP-418 in individuals with Limb-Girdle muscular dystrophy 2I, LGMDR9 FKRP-related (LGMD2I/R9)

Sproule D[1], *Reddy D*[1], *Blankenbiller T*[1], *Rainey A*[1], *Reklis L*[1]
[1]*ML Bio Solutions, A BridgeBio Company*

576P High precision quantification of ICAM-1 highlights similarities and differences between subgroups of Idiopathic inflammatory myopathies

Nishimura A[1], *Nelke C*[2], *Huber M*[3], *Mensch A*[4], *Roth A*[1], *Oberwittler C*[5], *Zimmerlein B*[6], *Krämer H*[7,8], *Neuen-Jacob E*[9], *Stenzel W*[10], *Müller-Ladner U*[3], *Ruck T*[2], *Schänzer A*[1]
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577P Evaluation of mitochondrial dysfunction as a potential mediator of inflammatory pathways in inclusion body myositis

Walli S[1], *Abdennebi D*[1], *Görg B*[2], *Bachir D*[1], *Dobelmann V*[1], *Preuße C*[3], *Stenzel W*[3], *Schoser B*[4], *Roos A*[1,6,7], *Brunn A*[5], *Meuth S*[1], *Ruck T*[1], *Nelke C*[1]
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578VP Recurrent rhabdomyolysis as the presenting feature of mixed connective tissue disease
Kent L[1], *Butterworth R*[2], *Hofer M*[1], *Giavri E*[2], *Brady S*[1]
[1]*Oxford University Hospitals NHS Foundation Trust,* [2]*Milton Keynes University Hospital*

644P-655P: Muscle MRI & new imaging techniques

644P Muscle MRI in Glycogen Storage disease type 5 (McArdle disease) – single centre experience
Mohamed K[1], *Pula S*[1], *Finnigan P*[1], *Godfrey R*[1], *Hammam A*[1], *Morrow J*[1], *Løkken N*[2], *Quinlivan R*[1]
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645P The utility and limitations of artificial intelligence in diagnosing rare disorders
Alawneh I[1], *Gonorazky H*[1], *Repalo A*[2], *Mashouri P*[2]
[1]*The Hospital for Sick Children,* [2]*University of Toronto*

646P Towards integration of quantitative MRI in the diagnostic protocol of neuromuscular disorders in three Dutch NMD expert centers
Rauh S[1], *Cameron D*[2], *Heskamp L*[3], *van Doorn J*[2], *Kruit M*[1], *Nievelstein R*[3], *Becks R*[2], *Niks E*[1], *van der Pol L*[3], *Houwen S*[2], *Braakman H*[2], *Erasmus C*[2], *van Alfen N*[2], *Bartels B*[3], *Froeling M*[3], *Kan H*[1]
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647P Tongue volume evaluation in Spinal and Bulbar Muscular Atrophy (SBMA) by AI-assisted automatic MRI-analysis
Rosenbohm A[1], *Vernikouskaya I*[2], *Müller H*[1], *Chadraabal K*[1], *Gadelkareem M*[1], *Ludolph A*[1,3], *Rasche V*[2,4], *Kassubek J*[1,3]
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648P MRI demonstrates altered skeletal muscle membrane permeability in Becker muscular dystrophy, representing a potential biomarker for disease activity
Schrama E[1], *Hooijmans M*[2], *Niks E*[1,3], *Kan H*[3,4], *Cameron D*[5]
[1]*Department of Neurology, Leiden University Medical Center,* [2]*Biomedical Engineering and Physics, Amsterdam University Medical Center,* [3]*Duchenne Center Netherlands,* [4]*C.J. Gorter MRI Center, Department of Radiology, Leiden University Medical Center,* [5]*Department of Medical Imaging, Radboud University Medical Center*

649P Comparing unsupervised AI techniques for visualizing MRI fat infiltration patterns in muscular dystrophies
Pizarro-Galleguillos B[1], *Goméz-Andrés D*[2], *Tobar F*[3], *Andia M*[4], *Díaz-Manera J*[5], *Verdú-Díaz J*[5], *Suazo Rojas L*[6], *Díaz Jara J*[6], *Bevilacqua J*[7]
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650VP Development and implementation of enhanced AI-derived DTI features for precision mapping of neural tract damage in Myotonic dystrophy
Kamali T[1], *Hageman N*[1], *Yazdavar T*[1], *C Piccolli*[1], *Day JW*[1], *Sampson J*[1], *Wozniak J*[2]
[1]*Department of Neurology and Neurological Sciences, Stanford University School of Medicine,* [2]*Department of Psychiatry & Behavioural Sciences, University of Minnesota*

651VP Quantitative MRI pre- and post-skeletal muscle biopsy reveals correlations with histopathological findings
Schlaffke L[1], *Forsting J*[1], *Kneifel M*[1], *Rehmann R*[1], *De Lorenzo A*[1], *Enax-Krumova E*[1], *Froeling M*[2], *Vorgerd M*[1,3], *Güttsches A*[1,3]
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652P Muscle MRI-phenotyping of patients with likely pathogenic anoctamin 5 variants

Poulsen N[1], *Palmio J*[2], *Jokela M*[2,3], *Claeys K*[4,5,15], *Iterbeke L*[5,15], *De Bleecker J*[6], *De Vos E*[6], *Domínguez-González C*[7], *Bermejo-Guerrero L*[7], *Muelas N*[8,9,10], *Vilchez J*[8,9], *Gonzalez-Chamorro A*[11], *Diaz-Manera J*[11], *Straub V*[11], *Straathof C*[12], *Niks E*[12], *Voermans N*[13], *Cameron D*[14], *Vissing J*[1], *On behalf of the study group*

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653P Rapid quantitative assessment of sodium dynamics after exercise using 23Na-MRI in dysferlinopathy and healthy controls

Bolano Diaz C[1], *Neal M*[2,3], *Richardson M*[1], *Muni Lofra R*[1], *Michell-Sodhi J*[1], *James M*[1], *Mayhew A*[1], *Hilsden H*[1], *Wilson I*[2,3], *Hollingsworth K*[2,3], *Blamire A*[2,3], *Straub V*[1], *Thelwall P*[2,3], *Diaz-Manera J*[1,4,5], ***Diaz-Manera J***[1,4,5]

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654P Different lower limb muscle MRI patterns in autosomal dominant titinopathies

Gomez Andres D[1], *Costa Comellas L*[1], *Díaz-Manera J*[2], *Õunap K*[3], *Álvarez-Molinero M*[1], *Urcuyo G*[1], *Savarese M*[4], *Munell F*[1], *Udd B*[5]

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655P Predictive modelling of dysferlinopathy progression: a longitudinal fat fraction analysis

Bolano Diaz C[1], *Wilson I*[2], *Hilsden H*[1], *James M*[1], *Araujo E*[3], *Reyngoudt H*[3], *Blamire A*[2], *Jain COS Consortium*[4], *Carlier P*[3], *Straub V*[1], *Diaz Manera J*[1]

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688P-704P: Therapies for NMD

688P Sleep and health-related quality of life in rare neuromuscular disorders

Fjermestad K[1], *Brørs E*

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689P Highlights and key learnings from 15 years of the TREAT-NMD Advisory Committee for Therapeutics (TACT)

Turner C[1], *Robertson L*[2], *Bushby K*[1], *Csimma C*[3], *De Luca A*[4], *Heslop E*[1], *Wells D*[5], *Aartsma-Rus A*[6], *Straub V*[1]

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690P The prospect of direct benefit in first-in-human gene therapy studies in minors - an ethical analysis

Pirson I[1], *Niks E*[1], *de Vries M*[1], *de Graeff N*[1]

[1]*Leids Universitair Medisch Centrum*

691P Substantial improvement of shoulder function with a new physiotherapy approach in children with LGMD, MD and FSHD

Pelsma M[1,2], *Ijspeert J*[2], *Braakman H*[1], *Houwen S*[1,2], *Hendrix- van der Stegen A*[1,2]

[1]*Radboud University Medical Centre-Amalia Children's Hospital,* [2]*Radboud University Hospital – Rehabilitation*

692P Determination of in vitro phenotype of Myofibrillar myopathy type 6 (MFM6), caused by the P209L mutation in the BAG3 gene, using patient-derived fibroblasts, and the role of autophagy stimulators in the cell recovery

Snoch W[1], *Rintz E*[1], *Węgrzyn G*[1]

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693P Proposed NIH core for advanced genetic therapies: spotlight on clinical development of AAV products for neuromuscular disorders

Todd J[1,2], *Reoma L*[2], *Nath A*[2], *Martin S*[2], *Brooks K*[2], *Ano S*[3], *Dukhanina O*[3], *Lawal T*[4], *Brooks P*[5], *Ottinger E*[5], *Lomash R*[5], *Stan R*[5], *Goldfeder L*[6], *Venditti C*[7], *Bönnemann C*[1]

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694P AAV-based gene replacement for SELENON-related myopathy

Moghadaszadeh B[1], *Troiano E*[1], *Barraza-Flores P*[1], *Lee W*[1], *Beggs A*[1]

[1]*Division of Genetics and Genomics, The Manton Center for Orphan Disease Research, Boston Children's Hospital, Harvard Medical School*

695P Unveiling the mosaic of quality of life in muscular dystrophy: an analysis of PROMIS survey data in adults

Dixon M[1], *Vordos K*[1], *Krikov S*[1], *Reeder M*[1], *Butterfield R*[1]

[1]*University Of Utah, Department of Pediatrics*

696P ADAPT-NMD: a hybrid II study exploring the feasibility of delivering, evaluating, and implementing a self-management programme for people with neuromuscular disorders at a specialist neuromuscular centre

Lee L[1], *Kulnik S*[2], *Curran G*[3], *Boaz A*[4], *Ramdharry G*[1]

[1]*University College London,* [2]*Ludwig Boltzmann Institute for Digital Health and Prevention,* [3]*University of Arkansas for Medical Sciences,* [4]*King's College London*

697P Clinicians' perspectives on a new self-management support programme for people living with neuromuscular disorders

Lee L[1], *Kulnik S*[2], *Boaz A*[3], *Ramdharry G*[1]

[1]*University College London,* [2]*Ludwig Boltzmann Institute for Digital Health and Prevention,* [3]*King's College London*

698P Adapting to life with a neuromuscular disorder: a qualitative exploration of the patient perspective

Lee L[1], *Kulnik S*[2], *Boaz A*[3], *Ramdharry G*[1]

[1]*University College London,* [2]*Ludwig Boltzmann Institute for Digital Health and Prevention,* [3]*King's College London*

699P Exon skipping for the second Calponin Homology Domain of dystrophin using AAV.U7snRNA - In vitro & Intramuscular studies using a novel murine model of Duchenne muscular dystrophy

Brinkman A[1], *Lesman D*[1], *Li D*[1], *Rajakumar D*[1], *Atre C*[1], *Rodriguez Y*[1], *Almeida C*[1], ***Wein N***[1]

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700P Alternative delivery of adeno-associated virus 9 for the treatment of Duchenne muscular dystrophy to target CSF and muscles– GFP Biodistribution study in WT mice

Iyer A[1], *Gomez C*[1], *Rodriguez Y*[1], *Almeida C*[1], *Bobbili P*[2], *McGovern V*[2], *Arnold D*[2], *Burghes A*[2], *Meyer K*[1], ***Wein N***[1]

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701P AAV.U7.ex44 Mediates Efficient Exon Skipping, Protein Restoration & Phenotype Rescue – Pre-Clinical Intramuscular and Dose Escalation Study for a Mutational Hotspot of the Duchenne Muscular Dystrophy (DMD)

Rajakumar D[1], *Almeida C*[1], *Atre C*[1], *Lesman D*[1], *Li D*[1], *Rodriguez Y*[1], *Young C*[2], *Spencer M*[2], *Flanigan K*[1], ***Wein N***[1]

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702P SEPN1/SELENON-related myopathy depends on the oxidoreductase ERO1A and is druggable with the chemical chaperone TUDCA

Ferreiro A[1,2], *Germani S*[3,4], *Tri Van Ho A*[1], *Cherubini A*[3], *Varone E*[3], *Chernorudskiy A*[3], *Renna G*[3], *Fumagalli S*[3], *Bolis M*[5], *Rastelli G*[6], *Nogara L*[7,8], *Poggio E*[9], *Brini M*[9,10], *Cattaneo A*[11], *Bachi A*[11], *Simmen T*[12], *Calì T*[7], *Boncompagni S*[6], *Blaauw B*[7,14], *Zito E*[3,4]

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703P Integrated Organ-on-Chip platform with PINP plasmonic biosensor for fibrosis monitoring in Duchenne muscular dystrophy

Fernández-Costa J[1], *Ruíz-Gutiérrez M*[1], *Ninfali C*[1], *Torabi M*[1], *Fernández-Simón E*[2], *Díaz-Manera J*[2], *Ramón-Azcón J*[1,3]

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704P Translating RNAi-based gene therapy for Facioscapulohumeral muscular dys-trophy

Wallace L[1], *Camp J*[1], *Neal K*[1], *Zender G*[1], *Taylor N*[1], *Zhou B*[1], *Ye F*[1], *Price B*[2], *Triplett M*[2], *Harper S*[1,3]

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15:30-16:00	**Short Oral Presentations 1** North Hall **146P, 145P, 231P, 576P, 577P** *Moderator: Werner Stenzel, Charité University Hospital*	**Short Oral Presentations 2** Terrace 2A **278P, 703P, 702P, 279P, 704P** *Moderator: Perry Shieh, University of California*	**Short Oral Presentations 3** Terrace 2B **400P, 399P, 654P, 655P, 653P** *Moderator: Krista Vandenborne, University of Florida*
16:15-17:00	**Debate: Can the costs of gene therapies in neuromuscular disorders be justified?** Congress Hall *Moderators: Francesco Muntoni, University College London, Great Ormond Street Hospital, UK & Teresinha Evangelista, Institut de Myologie, France*		
	14INV *Olga Germanenko, SMA Family Foundation, Russia* **15INV** *Josie Godfrey, JG Zebra Consulting, United Kingdom*		

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17:15-18:15	**Poster Session 2** Forum Hall (refreshments provided)

01P-51P, 52VP-54VP: CM - CMD

01P Structural variation in nebulin and its implications on phenotype and inheritance: establishing a dominant distal phenotype caused by large deletions

Sagath L[1,2], *Kiiski K*[1,3], *Naidu K*[4,5], *Djordjevic D*[6], *Yoon G*[6,7], *Rogers C*[8], *Scherer K*[9], *Koparir E*[10], *Kunstmann E*[11], *Davis M*[12], *Purwa J*[13], *Zygmunt A*[14], *Bönnemann C*[15,16], *Biancalana V*[17], *Echaniz-Laguna A*[18,19], *Beggs A*[20], *Henning F*[4], *Wallgren-Pettersson C*[1,2], *Pelin K*[1,2,21], *Lehtokari V*[1,2]

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02P Exploring genotype-phenotype correlations in NEB-related Myopathies

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

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03P Modulation of the cyclin inhibitor p27 can ameliorate muscular dystrophy in LAMA2-RD

Bonaccorso R[1], *Porrello E*[1], *Tonlorenzi R*[1], ***Previtali S***[1]

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04P Characterising patient iPSC-derived models of RYR1-related myopathies

Crane J[1,2], *Clayton J*[1,2], *Vo C*[1,2], *Driver K*[1,2], *Malfatti E*[3,4], *Romero N*[5,6], *Laing N*[1,2], *Ravenscroft G*[1,2], *Taylor R*[1,2]

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05P Pediatric cardiomyopathy may be more prevalent amongst ACTA1 mutation related myopathies: a single center case-series

Nandi D[1], *Meyer A*[2], *Connolly A*[3,4], *Waldrop M*[3,4], *Flanigan K*[3,4], *Cripe L*[1], *Hayes E*[1]

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06VP In vivo modulation of novel modifier genes for LAMA2-RD

Pini V[1,2], *Bonaccorso R*[1], *Porrello E*[1], *Burris T*[3], *Morgan J*[2], *Muntoni F*[2], *Previtali S*[1]

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07P Development of cell therapy for Ullrich congenital muscular dystrophy by iPSC-derived mesenchymal stromal cell

Sakurai H[1], *Goto M*[1], *Takenaka-Ninagawa N*[1], *Harada A*[1], *Ikeya M*[1]

[1]*Center for iPS Cell Research and Application (CiRA), Kyoto University*

08P SRPK3-TTN related myopathy: early clinical characteristics and muscle imaging findings

Orbach R[1], *Donkervoort S*[1], *Saade D*[2], *D'Souza P*[3], *Haugland J*[1], *Foley A*[1], *Bharucha Goebel D*[1,4], *Potticary A*[1], *Chao K*[5], *Macnamara E*[3], *Finkel R*[6], *Beggs A*[7], *Tifft C*[3], *Bönnemann C*[1]

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09P Delineating collagen VI genes expression patterns in mice skeletal muscles using RNAscope

Guirguis F[1,2], *Bolduc V*[1], *Zhou H*[3,4], *Muntoni F*[2,3], *Bönnemann C*[1]

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10P Characterization of the severe phenotype of COL6-related dystrophy due to the recurrent deep intronic pseudoexon-inducing variant COL6A1 c.930+189C>T

Foley A[1], *Bolduc V*[1], *Guirguis F*[1], *Donkervoort S*[1], *Hu Y*[1], *Orbach R*[1], *Mohassel P*[1], *Zhou H*[2], *Aguti S*[3], *Jimenez-Mallebrera C*[4], *Lamandé S*[5], *Allamand V*[6], *Gualandi F*[7], *Ferlini A*[7], *Wilton S*[8,9], *Wagener R*[10], *Bertini E*[11], *Muntoni F*[12,13], *Bönnemann C*[1], *The COL6A1 Intron 11 Study Group*

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11P Autosomal dominant centronuclear myopathy caused by variants in the DNM2 gene – Results of an international, prospective natural history study

Braun F[1,2], *Seferian A*[3], *Behin A*[4], *Fer F*[3], *Peretti M*[3], *Duchêne D*[3], *Schara-Schmidt U*[1], *Annoussamy M*[3], *Baets J*[5,6], *Servais L*[3], *NatHis-CNM Study Group*[7]

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12P Female carriers of X-linked Myotubular Myopathy (XL-MTM) in Germany – extending the knowledge about the impact of heterozygous variants in the MTM1 gene

Braun F[1,2], *Kaiser F*[2], *Schara-Schmidt U*[1]

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13P Centronuclear Myopathy in DNM2 E368K mice: behavioral and pathological insights

Uy G[1], *Ogawa M*[3], *Inoue Y*[2], *Takeda T*[3], *Inoue T*[2], *Hayashi S*[1], *Nishino I*[1], *Noguchi S*[1]

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14P X-linked myotubular myopathy: 3-year follow-up of a prospective international natural history

Seferian A[1], *Annoussamy M*[2], *Fer F*[2], *Lilien C*[1,3], *Gidaro T*[1], *Schara-Schmidt U*[4], *Braun F*[4,5], *D'Amico A*[6], *Daron A*[7], *Hernández González A*[8], *de Lattre C*[9,10], *Villerot C*[11], *Behin A*[12], *Arnal J*[13], *Mayer M*[14], *Bellance R*[15], *Davion J*[16], *Hogrel J*[17], *Servais L*[1], *on behalf of NatHis CNM Study Group*

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15P Prospective, longitudinal study of the natural history of paediatric patients in France with LAMA2 related dystrophies

Seferian A[1], *Gasnier E*[2], *Peretti M*[2], *Duchene D*[2], *Lagrue E*[2], *Vuillerot C*[3], *Walther-Louvier U*[4], *Gómez García-de-la-Banda M*[5], *Grange A*[2], *Carlier R*[6], *Reyngoudt H*[7], *Marty B*[7], *Colella M*[1], *Quijano-Roy S*[5]

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16P Limitation of short-reads NGS sequencing on genomic DNA: interest of functional studies in the diagnosis of congenital Ullrich muscular dystrophy

Metay C[1,2], *Ghanem R*[1], *Toutain A*[3], *Bloch A*[1], *Blin E*[1], *Jobic V*[1], *Pham T*[4], *Lejeune E*[5], *Buratti J*[5], *Keren B*[5], *Ader F*[1,6,7,8], *Richard P*[1,6,7]

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17P Decoding TTN gene mutations in Indian cohort: detection and characterization using insights from sequencing and structural analysis

Dhall A[1], *Jassal B*[1], *Faruq M*[2], *Shamim U*[2], *Kumar D*[1], *Bhatia D*[1], *Venugopalan Y V* [1], *Suri D*[1], *Sharma D*[1]

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18P Cohort of Czech patients with RYR1-related disorders

Lauerová B[1], *Zidkova J*[2,3], *Rohlenova M*[1], *Dolanska A*[1], *Gloser M*[1], *Kumhera M*[1], *Lassuthova P*[4], *Kramarova T*[2,3], *Kopcilova J*[2], *Fajkusova L*[2,3], *Haberlova J*[1]

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19P Recurrent TTN missense variants in biallelic titinopathies: collecting all the puzzle pieces

Di Feo M[1,2], *Rees M*[3], *Gautel M*[3], *Jungbluth H*[3,4], *Fiorillo C*[1,5], *Bruno C*[5,6], *Udd B*[2,6], *Savarese M*[2], *On behalf of the Titin Study Group*[7]

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20P A novel DNM2 variant causing Centronuclear Myopathy with respiratory involvement

Urra A[1], *Fajre F*[1], *Pizarro B*[2], *Gonzalez D*[3], *Maquilon C*[4], *Antolini M*[4], *Bevilacqua J*[1,5]

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21P Tongue ultrasound assessment in patients with Nemaline myopathy

Moreno C[1], *Moreira A*[1], *Gontijo Camelo C*[1], *Zanoteli E*[1]

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22P Clinical and ultrasonographic evaluation of dysphagia in patients with LAMA2-CMD

Camelo C[1], *Sampaio P*[1], *Moreira A*[1], *Moreno C*[1], *Artilheiro M*[1], *Silva A*[1], *Reed U*[1], *Zanoteli E*[1]

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23P Establishing the genetic diagnosis in patients with suspected recessive Titinopathy

Mueller J[1], *Savarese M*[2], *Lillback V*[2], *Perry L*[1], *Zaharieva I*[1], *Pini V*[1], *Sagath L*[3], *Steyaert W*[3], *Hoischen A*[3], *Yepez V*[4], *Esteve-Codina A*[5], *Neveling K*[3], *Topf A*[6], *Muntoni F*[1], *Sarkozy A*[1]

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24P Improving gross motor function in pediatric Centronuclear Myopathy with acetylcholinesterase inhibitors: a case report

Paprad T[1], *Laohapongsomboon S*[1], *Amornvit J*[2]

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25P Data trends and highlights from The Global Registry for COL6-related Dystrophies

McDonald S[1], *Allamand V*[2], *Alvarez R*[3], *Dziewczapolski G*[3], *Boddy H*[4], *Deconinck N*[5], *Ferré X*[6], *McAlister B*[4], *Mejat A*[7], *Sarkozy A*[8], *Copier J*[9], *Straub V*[1]

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26P Is oral salbutamol a possible treatment for congenital myopathies?

Michael E[1,2], *Hedberg-Oldfors C*[3], *Gudmundsson M*[4], *Weichbrodt J*[4], *Oldfors A*[3], *Darin N*[2]

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27P Muscle ultrasound findings in paediatric patients with TTN gene related congenital myopathy

Sarkozy A[1], *Perry L*[1], *Cicala G*[1,2], *Manzur A*[1], *Vanegas M*[3], *Khries M*[3], *Sudhakar S*[4], *Clark C*[5], *Muntoni F*[1,6], *Jungbluth H*[3], *Baranello G*[1]

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28P CHKB muscular dystrophy: beyond Megamitochondria

Codina Bergada A[1,2], *Nascimento A*[1], *Lavilla R*[2], *Ortez C*[1], *Natera-de Benito D*[1,3], *Expósito J*[1], *Estévez-Arias B*[1,4], *Carrera L*[1], *Martorell L*[5], *Isaí A*[1], *Colomer J*[1], *Jou C*[1,2,3,6]

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29P Temporal requirement of dystroglycan glycosylation during brain development and rescue of cortical dysplasia via gene delivery in the fetal stage

Sudo A[1], *Kanagawa M*[2], *Kobayashi K*[2], *Endo M*[3], *Minami Y*[3], *Aiba A*[4], *Toda T*[1]

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30P Nemaline Myopathy-linked TNNT1 mutations are associated with aberrant thin filament extensibility and myofibre hyper-contractility

Laitila J[1,2], *Lewis C*[2], *Hessel A*[3,4], *Primiano G*[5,6], *Hernandez-Lain A*[7,8], *Fiorillo C*[9], *Lawlor M*[10], *Ottenheijm C*[11], *Ochala J*[2]

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31P Nutritional status of patients with nemaline myopathy and related congenital myopathies in Finland

Lehtokari V[1,2], *Similä M*[3], *Tammepuu M*[4], *Isohanni P*[5], *Auranen M*[6], *Hiekkala S*[7], *Wallgren-Pettersson C*[1,2], *Strang-Karlsson S*[8]

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32P Investigating myosin dysregulation in X-linked myotubular myopathy

Rostedt F[1,2], *Gerlach Melhedegaard E*[1], *Zanoteli E*[3], *Primiano G*[4], *Nishino I*[5], *Laporte J*[6], *Gineste C*[6], *Romero N*[7], *Lawlor M*[8], *Wallgren-Pettersson C*[2,9], *Laitila J*[1,2], *Ochala J*[1]

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33P Description of natural history baseline characteristics of a paediatric cohort of recessive TTN myopathy patients in the UK – a prospective study

Wolfe A[1,2], *Cicala G*[1,2], *Joefield T*[2], *Sheehan J*[3], *Main M*[1], *Vanegas M*[3], *Khries M*[3], *Clark C*[1,2], *Sudhakar S*[1], *Perry L*[1], *Bilby J*[1], *Jungbluth H*[3], *Muntoni F*[1,2,4], *Baranello G*[1,2,4], *Sarkozy A*[1,2]

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34P Investigating sarcomeric changes in Cullin-3 knockout muscles

Fochi V[1], *Blondelle J*[2], *Medelyte B*[1], *Seto J*[3,4], *Jones Y*[2], *Castillon G*[2], *Ghassemian M*[5], *Singer J*[6], *Lange S*[1,2,7]

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35P Generation of a zebrafish knock-in model of Exertional Heat Stroke

Endo Y[1,2], *Geissah S*[2], *Cui X*[2], *Noche R*[2], *Dowling J*[2]

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36P Liver involvement in Myotubular and Centronuclear Myopathy: review of one year's data collected by the MTM & CNM Patient Registry

Bohill J[1], *Ward E*[2], *Lennox A*[3], *Lawlor M*[4], *Jungbluth H*[5,6], *Beggs A*[7], *Graham R*[8], *Heidemann M*[9], *Wood M*[2], *Ward M*[2], *Page J*[1], *Cowling B*[10], *Haselkorn T*[11], *Voermans N*[12], *Foley A*[13], *Kyrana E*[14], *Marini-Bettolo C*[1], *Dhawan A*[14], *Dowling J*[15]

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37P TreatMYPN: In vivo mouse characterization and in vitro 3D muscle studies followed by 15P AAV gene therapy for the ultrarare myopalladin-related Congenital Myopathy

Onnée M[1], *Taglietti V*[1], *Gicquel E*[2], *Bastu S*[1], *Periou B*[1,3], *Banos G*[1], *Mignot J*[1], *Bleuzen A*[1], *Roudaut C*[2], *Tiret L*[1], *Didier N*[1], *Richard I*[2], *Relaix F*[1], *Malfatti E*[1,3]

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38P Collablot, a new immunodetection assay for the quantification of collagen VI expression and secretion in cell culture

Sendino M[1], *Martín-Gonzalez S*[1], *Osegui-Barcenilla N*[1], *López-Márquez A*[2], *González-Moro I*[1], *Benito-Agustino A*[1], *López-Martínez A*[1], *Jimenez-Mallebrera C*[2], ***Arechavala Gomeza V***[1,3]

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39P Studying markers of metabolism, apoptosis, and necrosis in cullin-3 skeletal muscle knockouts

Herskind J[1], *Fujita K*[2], *Dato V*[2], *Fochi V*[1], *Blondelle J*[2], *Medelyte B*[1], *Seto J*[3,4], *Jones Y*[2], *Castillon G*[2], *Ghassemian M*[5], *Singer J*[6], *Lange S*[1,2,7]

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40P Muscle ultrasound pattern in Merosin-deficient muscular dystrophy

Costa Comellas L[1,2,3], *Gómez-Andrés D*[1,2,3], *Urcuyo G*[1,3], *Toro-Tamargo E*[1], *López-López J*[4], *Álvarez-Molinero M*[1,5], *Munell F*[1,2,3]

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41P Rigid spine syndrome revealing nemaline myopathy caused by a novel mutation in cofilin-2 gene (CFL2)

Farhat E[1,3], *Miladi N*[1,4], *Chaabouni M*[1,2]

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42P Characterizing MRI brain abnormalities in X-linked Myotubular Myopathy

Vogt L[1], ***Amburgey K***[1], *Widjaja E*[2], *Dowling J*[1]

[1]*Hospital For Sick Children,* [2]*Lurie Children's Hospital*

43P Characterization of RYR1 variants in congenital myopathy zebrafish models

Sinclair J[1], *Todd J*[1], *Feldman B*[1], *Lawal T*[1]

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44P Novel TTN mutation causing a congenital onset, slowly progressive myopathy with contractures in two siblings

Figueroa Bonaparte S[1], *Martinez -Viguera A*[1], *Juanola-Mayos E*[1], *Lucente G*[1], *Almendrote M*[1], *Gasch-Navalon E*[2], *Corral-Juan M*[2], *Matilla-Dueñas A*[2], *Martinez-Piñeiro A*[1]

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45P Strategies to improve the design of gapmer antisense oligonucleotide on allele-specific silencing in COL6-related congenital muscular dystrophies

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46P Congenital Myopathies and Muscular Dystrophies in the UK: a comprehensive next generation sequencing analysis of frequencies over an 8-year period (2016-2023)

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47P MYBPC1 mutation in a family: spectrum of phenotype in three generations

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48P Digging into histological-genetic correlations in MYH2-myopathy: a case series and review of the literature

***Labella B**[1,2], Brochier G[1,3], Beuvin M[1], Méneret A[3], Leonard-Louis S[3], Maisonobe T[3], Stojkovic T[1], Métay C[3], Evangelista T[1,3]*
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49P Epidemiologic study of XLMTM and clinical expression in the liver (EXCEL): study design of an observational, patient-centric study

***Beggs A**[1], **Haselkorn T**[2], Dhawan A[3], Lawlor M[4], Kim J[2], Ward E[5], Hughes Z[6], Baima J[2], James L[2], Coats J[2], Gentyala R[2], Brandon T[2], Jesus Perez Inigo Garcia-Malo M[7], Marini Bettolo C[8], Graham R[9], Dowling J[10]*
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50P Monoallelic DAG1 truncating variants in patients with hyperCKemia

***Segarra-casas A**[1,2], Trainor C[1], Polavarapu K[3], Díaz-Manera J[1], Gonzalez-Quereda L[2,4], Kirschner J[5], López de Munain A[6,7,8,9], Nascimento A[4,10,11], Roos A[3,12,13], Dowling J[14,15], Muntoni F[16,17], DAG1 Study Group, Töpf A[1], Straub V[1]*
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51P Clinical characterization of Collagen XII-related disease caused by biallelic COL12A1 variants

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52VP Muscle ultrasound as a potential biomarker in patients with LAMA2-related dystrophy under 6 years of age

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53VP A 5-year natural history study in LAMA2-related muscular dystrophy and SELENON-related myopathy: the extended last strong study

De Laat I[1], *Bouman K*[1,2], *van de Camp S*[1], *Dittrich A*[3], *van Tilburg W*[4], *Cameron D*[5], *Doorduin J*[1], *van Alfen N*[1], *Kamsteeg E*[6], *Houwen S*[7], *Groothuis J*[7], *Erasmus C*[2], *Voermans N*[1]
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54VP Emerging muscle ultrasound patterns as a diagnostic aid in TTN-related myopathy

Potticary A[1], *McAnally M*[1], *Donkervoort S*[1], *Bönnemann C*[1]
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79P-97P: LGMD

79P Profiling of pathogenic variants in Japanese patients with Sarcoglycanopathy

Shimazaki R[1], *Saito Y*[1,2], *Awaya T*[3,4], *Minami N*[2], *Kurosawa R*[3], *Hosokawa M*[3], *Ohara H*[3], *Hayashi S*[1], *Takeuchi A*[5], *Hagiwara M*[3], *K. Hayashi Y*[6], *Noguchi S*[1], *Nishino I*[1,2]
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80P Evaluating the clinical significance of single heterozygous likely pathogenic and pathogenic variants for autosomal recessive limb-girdle muscular dystrophies in a highly specialised service for rare neuromuscular disorders

Oliwa A[1], *Hudson J*[1], *Topf A*[1], *Graham E*[1], *Salman D*[1], *Straub V*[1], *Harris E*[1], *Marini-Bettolo C*[1]
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81P Minimal clinically important differences in Dysferlinopathy from the 10-year, multicenter Jain Clinical Outcome Study

Gordish-Dressman H[1,2], *James M*[3], *Rufibach L*[4], *Hilsden H*[3], *Day J*[5], *Mendell J*[6], *Fernandez-Torron R*[7], *Harms M*[8], *Pestronk A*[9], *Vissing J*[10], *Desai U*[11], *Yoshimura M*[12], *Shin J*[13], *Mozaffar T*[14], *Stojkovic T*[15], *Pegoraro E*[16], *Bevilacqua Rivas J*[17], *Olive M*[18], *Paradas C*[19], *COS consortium on behalf of the Jain Foundation, Straub V*[3]
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82P Diagnostic approach to ANO5-Related Muscular Dystrophy in a series of female siblings: an interdisciplinary perspective

Correa Arrieta C[1], *Castellar Leones S*[1,2,3], *Calderon Castro A*[1,2,3], *Rodriguez P*[1,2], *Saldaña D*[1], *Maradei Anaya S*[1,2]
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83P Should we include HACD1 variants in the differential diagnosis of slowly progressive adult-onset limb-girdle weakness?

Da Silveira Massaro C[1], *Ferreira W*[1], *Farias I*[1], *Oliveira H*[1], *Serrano P*[1], *Badia B*[1], *Souza P*[1], *Bezerra M*[1], *Pinto W*[1], *Nunes K*[1], *Oliveira A*[1]
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84P A rare homozygous CAPN3 variant with distinct clinical features in unrelated families of Iraqi Jewish descent

Aharoni S[2,4], *Assia Batzir N*[1], *Orenstein N*[1,2], *Yaron Y*[2,3], *Kuzminsky A*[4], *Nevo Y*[2,4], *Konen O*[2,5], *Bazak L*, *Lidzbarsky G*[6], *Basel-Salmon L*[2,6,7]

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85P Myopathy secondary to SCN4A C.3502C>T variant

Calabria Gallego M[1]

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86P Muscle 3D models to investigate LGMDD2 Transportin 3 Related Deficiency: insights into myogenic processes and contractile dysfunction

Pacilio S[1,4], *Costa R*[1], *Rodia M*[1], *Lombardi S*[1], *Di L*[2], *Banos G*[3], *Didier N*[3], *Malfatti E*[4], *Focarete M*[2], *Cenacchi G*[1]

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87P MRI characterization of the cardiac involvement in LGMD2i/R9

Fromes Y[1], *Olivier S*[2], *Zanfongnon R*[3], *Thevenot E*[3], *Stojkovic T*[1,4], *Marty B*[1], *Reyngoudt H*[1]

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88P Clinical progression of 43 patients with Sarcoglycanopathy at a single centre

Kocak G[1], *Michel-Sodhi J*[1], *Marini-Bettolo C*[1], *Moat D*[1], *Guglieri M*[1], *Robinson E*[1], *Schiava M*[1], *Bolano-Díaz C*[1], *Wong K*[1], *Grover E*[1], *James M*[1], *Segovia S*[1], *Elseed M*[1], *Salman D*[1], *Harris E*[1], *Topf A*[1], *Luce L*[1], *Hudson J*[2], *Henderson M*[2], *Parkhurst Y*[2], *Graham E*[2], *Straub V*[1], *Tasca G*[1], *Muni Lofra R*[1], *Diaz-Manera J*[1]

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89P The LGMD2A/Calpainopathy Registry: A patient-powered natural history study and trial recruitment tool

Levy J[1], *Boslego J*[1], *Guglieri M*[2], *Martin A*[3], *Mathews K*[4], *Wrubel M*[1]

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90P Genetic heterogeneity of limb girdle myopathies in Tunisia: more than sarcoglycanopathies

Farhat E[1], *Miladi N*[1], *Chaabouni M*[1,2], *Amouri R*[3], *Leturcq F*[4]

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91P Determination of gene/protein expression and protein functionality in carriers of Limb-Girdle Muscular Dystrophy (LGMD)

Gaynor A[1]

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92P Development and characterization of induced pluripotent stem cells derived from Calpainopathy patients, carrying the CAPN3 c.1746-20C>G variant

Inashkina I[1], *Goluba K*[2], *Tvoronoviča A*[1], *Parfejevs V*[2], *Lāce B*[2,3], *Riekstiņa U*[2]

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93P The International Clinical Outcome Study for Dysferlinopathy II: validation of motor outcome measures in a new patient cohort

Hilsden H[1], *James M*[1], *Gordish Dressman H*[2], *Rufibach L*[3], *Day J*[4], *Mendell J*[5], *Fernandez Torron R*[6], *Harms M*[7], *Pestronk A*[8], *Vissing J*[9], *Desai U*[10], *Yoshimura M*[11], *Shin J*[12], *Mozaffar T*[13], *Stojkovic T*[14], *Pegoraro E*[15], *Bevilacqua Rivas J*[16], *Olive M*[17], *Paradas C*[18], *Straub V*[1]

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94P JOURNEY: a natural history study of Limb Girdle muscular dystrophies R3–R5: baseline characteristics of study cohort

Lowes L[1], ***Ortez Gonzalez C***[2], *Claeys K*[3], *Laverty C*[4], *Gangfuss A*[5], *Proud C*[6], *Manera J*[7], *James M*[7], *Alfano L*[1], *Stevenson H*[8], *Yu L*[8], *Comi G*[9,10]

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95P Development of a myotropic gene therapy towards a first treatment for LGMDR7

Heier C[1], *Fiorillo A*[1], *Provenzano M*[1], *McMichael G*[1], *Crumbaugh C*[1], *Hawkins E*[1], *Johnson N*[1], *Hale M*[1]

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96P The impact of losing the ability to sit to stand on social participation in people with Dysferlinopathy: clinical outcome study for Dysferlinopathy

Robinson E[1], *James M*[1], *Hilsden H*[1], *Rufibach L*[2], *Roper W*[3], *Holsten S*[4], *Lowes L*[5], *De Monts C*[6], *Yochai C*[7], *Zabala Pardo A*[8], *Ogasawara Y*[9], *Rudolph K*[10], *Weber J*[11], *Montiel Morillo E*[12], *Birnbaum S*[13], *Rojas Rojas J*[14], *Mayhew A*[1], *Straub V*[1], *COS Consortium on behalf of the Jain Foundation*[3]

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97P Spectrum of limb girdle muscular dystrophies in a cohort of inherited myopathies with limb girdle weakness

Venugopalan Y V[1], *Macken W*[2,3], *Dalal A*[4], *Rani N*[1], ***Reyaz A***[1], *Ahmad T*[1], *Tarane K*[1], *Danish M*[1], *ICGNMD Consortium*[5], *Bhatia R*[1], *Wilson L*[2], *Bugiardini E*[2], *Vandrovcova J*[2], *Houlden H*[2], *Pitceathly R*[2,3], *Thangaraj K*[4,6], *Topf A*[7], *Straub V*[7], *Hanna M*[2], *Srivastava P*[1]

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401P-436P, 437VP: DMD – treatments

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401P Moderate-term dimethyl fumarate treatment reduces fibrosis of skeletal and cardiac muscle in in the mdx mouse model of Duchenne muscular dystrophy

Timpani C[1,2,3], Kourakis S[1,2], Bagaric R[1,2], Qi B1[2], Ali B[1,2], Spiesberger G[1,2], Boyer R[1], Kandhari N[4], Yan X[1,5], Stupka N[3,2,1], van Putten M[6], Aartsma-Rus A[6], Deveson-Lucas D[4], Fischer D[7], Rybalka E[1,2,3]

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402P Faecal microbiota transplantation: a potential novel therapeutic avenue for Duchenne muscular dystrophy

Timpani C[1,2,3], Debrincat D[1], Van Der Beek J[1], Eri R[4,5], Diwakarla S[6,3], McQuade R[6,3,2], Rybalka E[1,2,3]

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403P CONNECT1-EDO51: A 12-week open-label Phase 2 study to evaluate PGN-EDO51 safety and efficacy in people with Duchenne amenable to exon 51 skipping

Mellion M[1], McMillan H[2], Chrestian N[3], Gonorazky H[4], O'Connell C[5], Vacca S[1], Peterson M[1], Marcos B[1], Batra S[1], Lonkar P[1], Holland A[1], Foy J[1], Lamore S[1], Garg B[1], Yu S[1], Larkindale J[1]

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404P CONNECT2-EDO51: A Phase 2 placebo-controlled study to evaluate PGN-EDO51 safety and efficacy in people with Duchenne amenable to exon 51 skipping

Mellion M[1], Marcos B[1], Vacca S[1], Peterson M[1], Batra S[1], Lonkar P[1], Holland A[1], Foy J[1], Lamore S[1], Garg B[1], Yu S[1], Larkindale J1

[1]Pepgen Inc.

405P Single- and repeat-dose nonclinical data for PGN-EDO51 demonstrated favorable pharmacology and safety profiles for the treatment of DMD

Sweeney C[1], Gilbert J[1], Lamore S[1], Lonkar P[1], Foy J[1], Holland A[1]

[1]PepGen Inc.

406P Effects of telerehabilitation-based motor imagery training on motor imagery ability and motor function in children with Duchenne muscular dystrophy

Bora Zereyak M[1], Bulut N[1], Yılmaz Ö[1], Gürbüz İ[1], Haliloğlu G[2]

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407P RGX-202, an investigational gene therapy for the treatment of Duchenne muscular dystrophy: interim clinical data

Dastgir J[1], Veerapandiyan A[2], Rao V[3], Tesi-Rocha C[4], Harper A, Iannacone S[6], Falabella P[1], Pakola S[1], Philips D[1], Wilson C[1], Boulos N[1], Gilmor M[1], Yang L[1], Patel H[1], Fiscella M[1], Danos O[1]

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408P Dose escalation and repeatability for ultrasound-guided intramuscular administration of local treatment in Duchenne muscular dystrophy

Findik Sener O[1], Sage F[1], Niks E[1,2], de Ruiter M[1], Engelse M[1], Geijsen N[1], Burgmans M[1], Kan H[1,2]

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409P Short-term and long-term safety profile of Givinostat in Duchenne muscular dystrophy

Vučinić D[1], Nascimento A[2], Finkel R[3], Brandsema J[4], Finanger E[5], Harper A[6], Acsadi G[7], Nevo Y[8], Houwen-van Opstal S[9], Blaschek A[10], Coceani N[11], Cazzaniga S[11], Bettica P[11], Muelas N[12]

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410P GNT0004, Genethon's AAV8 vector-delivered microdystrophin gene therapy of Duchenne muscular dystrophy, first data of the phase I/II part of the GNT-016-MDYF all-in-one clinical trial in ambulant boys

*Laugel V[1], De Lucia S[2], Davion J[3], Daniele N[4], **Cao F**[4], Sanz M[4], Buscara L[4], Blaie S[4], Thibaut L[4], Sagot M[4], Riviere A[4], Creoff E[4], Lelait M[4], Valent A[4], Perret G[4], Braun S[4], Muntoni F[5]*

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411P Phenotypical differences in 2 immortalized dystrophic cell lines from pediatric patients: a glimpse into pharmacological personalized therapy

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412P Duchenne muscular dystrophy fibro-adipogenic progenitors impair muscle function of co-cultured healthy myotubes in a functional 3D model

***Ninfali C**[1], Fernández-Garibay X[1], Tejedera-Villafranca A[1], Díaz-Manera J[2], Ramón-Azcón J[1,3], Fernández-Costa J[1]*

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413P Natural history of renal dysfunction in Duchenne muscular dystrophy

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414P Pulmonary function in Colombian non-ambulatory patients with Duchenne muscular dystrophy treated with Ataluren

***Gordillo Gonzalez G**[1,2], Villamil-Osorio M[3], Restrepo-Gualteros S[4], Vallejo-Mesa D[5,6], Rivera-Nieto C[7], Contreras-García G[8,9], Guerra-Araujo V[10], Ospina-Lagos S[11,12], Paredes-Brijaldo Á[4], Hernández-Forero M[13], Becerra-Ortíz P[14], Silvera-Redondo C[15], Arias C[16], Campo-Ternera C[17,18], Curiel-Acosta J[19]*

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415P Cardiac function in Colombian non-ambulatory patients with Duchenne muscular dystrophy treated with Ataluren

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416P Effect of ataluren on upper limb function in nmDMD patients from the STRIDE Registry

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417P Comparison of changes in 6-minute walk distance for Colombian patients with Duchenne muscular dystrophy treated with Ataluren with different rates of decline

Ruiz Ospina E[1,2], *Castellar Leones S*[1,3], *Bobadilla Quesada E*[2], *Bolaños Almeida C*[2], *Ladino Cortes L*[2], *Ruiz Arbelaez C*[2], *Maradei Anaya S*[2], *Parra Dugarte N*[4], *Contreras García G*[5,6], *Ramírez Rodríguez S*[7,8], *Gomez Castillo C*[9], *Ospina Lagos S*[1], *Sierra del Villar G*[4], *Becerra Ortiz P*[10], *Silvera Redondo C*[11], *Sierra Rosales G*[12]
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418P Ambulatory function in Colombian patients with Duchenne muscular dystrophy treated with Ataluren after a baseline 6-minute walk distance <300m

Ruiz Ospina E[1,2], *Castellar Leones S*[1,3], *Vallejo Mesa D*[4,5], *Contreras García G*[6,7], *Acosta Aragón M*[8], *Ospina Lagos S*[1], *Guerra Araújo V*[9], *Becerra Ortíz P*[10], *Silvera Redondo C*[11], *Campo Ternera C*[12,13], *Gomez Castillo C*[5], *Curiel Acosta J*[14], *Ramírez Rodríguez S*[15,16]
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419P A stapled inhibitory core peptide of myostatin improves dystrophic pathology in DBA/2-mdx mice

Sunada Y[1], *Nishimatsu S*[1], *Fujino M*[1], *Ohsawa Y*[1]
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420P Potential benefits of JMV2894, a growth hormone secretagogue, in Duchenne muscular dystrophy: insights from a preclinical study in the D2-mdx mouse

Mantuano P[1], *Boccanegra B*[1], *Cappellari O*[1], *Mele A*[1], *Marinelli M*[1], *Cristiano E*[1], *Tulimiero L*[1], *Conte E*[1], *Torsello A*[2], *Denoyelle S*[3], *Liantonio A*[1], *De Luca A*[1]
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421P Early therapeutic benefit of delandistrogene-moxeparvovec-rokl in Duchenne muscular dystrophy

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422P U7snRNA-mediated exon 17 skipping restores dystrophin expression in cells and in a novel mouse model of Duchenne muscular dystrophy

Gushchina L[1], *Dufresne G*[1], *Saylam E*[1], *Bradley A*[1], *Rohan N*[1], *Lowery A*[1], *Suhaiba A*[1], *Lin H*[1], *Wein N*[1], *Flanigan K*[1]
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423P Safety and efficacy of delandistrogene moxeparvovec versus placebo in Duchenne muscular dystrophy: phase 3 EMBARK primary results

Mendell J[1,2], *Muntoni F*[3], *McDonald C*[4], *Mercuri E*[5], *Ciafaloni E*[6], *Komaki H*[7], *Leon-Astudillo C*[8], ***Nascimento A***[9], *Proud C*[10], *Schara-Schmidt U*[11], *Veerapandiyan A*[12], *Zaidman C*[13], *Murphy A*[14], *Reid C*[14], *Asher D*[15], *Darton E*[15], *Mason S*[15], *Fontoura P*[16], *Elkins J*[15], *Rodino-Klapac L, on behalf of the EMBARK Study Group*[15]
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424P Micro-dystrophin expression and safety with delandistrogene moxeparvovec gene therapy for DMD in a broad population: phase 1B trial (ENDEAVOR)

Proud C[1], *Zaidman C*[2], *McDonald C*[3], *Day J*[4], *Thrasher P*[5], *Asher D*[5], *Murphy A*[6], *Guridi M*[7], *Ding K*[5], *Reid C*[6], *Lewis S*[5], *Magistrado-Coxen P*[5], *Palatinsky E*[5], *Wandel C*[7], *Potter R*[5], *Rodino-Klapac L*[5], *Mendell J*[8,9]

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425P Five-year outcomes with delandistrogene moxeparvovec in patients with Duchenne muscular dystrophy (DMD): a phase 1/2a study

Mendell J[1,2], *Sahenk Z*[1,2], *Lowes L*[1,2], *Reash N*[1], *Iammarino M*[1], *Alfano L*[1], *Signorovitch J*[3], *Jin J*[4], *Gao P*[4], ***Mason S***[4], *Elkins J*[4], *Rodino-Klapac L*[4]

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426P Ankle foot orthosis prescription for Duchenne muscular dystrophy patients: a retrospective study

Prufer De Q. C. Araujo A[1], *Ferreira Rebel M*[1], *Almeida Pereira J*[1], *Mello Brasil L*[1], *Chacon Pereira A*[1], *Horokoski Kovacs A*[1], *Pereira Dias Drumond Fortes C*[1], *Nardes dos Santos F*[1], *de Fatima Landgraaf J*[1], *Stajnbock F*[1]

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427P An open-label study to collect long-term safety and efficacy data from boys with DMD who have completed prior studies with vamorolone (GUARDIAN Study)

Nip K[1], *de Vera A*[1], *Hasham S*[1], *Charef P*[1], *Wong S*

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428P Cardiac MRI outcomes in patients with Duchenne muscular dystrophy treated with delandistrogene moxeparvovec: findings from EMBARK Part 1

Walter G[1], *Vandenborne K*[2], *Bourke J*[3], *Soslow J*[4], *Mason S*[5], *Palatinsky E*[5], *Wandel C*[6], *Ding K*[5], *Reid C*[7], *Murphy A*[7], *Manfrini M*[6], *Richardson J*[5], *Elkins J*[5]

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429P Givinostat in Duchenne muscular dystrophy: natural history comparison applying propensity score matching

Gomez Andres D[1], *Sansone V*[2], *Phan H*[3], *Willis T*[4], *Guglieri M*[5], *Scoto M*[6], *Vandenborne K*[7], *Cazzaniga S*[8], *Coceani N*[8], *Bettica P*[8], *McDonald C*[9]

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430P The damaged lysosome is a therapeutic target for combined therapy in Duchenne muscular dystrophy

Jaber A[1,2], *Bakour R*[1,2], *Palmieri L*[1,2], *Bourg N*[1,2], *Albini S*[1,2], *Richard I*[1,2], *Israeli D*[1,2]

[1]*Genethon,* [2]*Paris-Saclay University, Inserm, Integrare Research Unit UMR_S951*

431P Exon 45 skipping and dystrophin production with ENTR-601-45 in preclinical models of Duchenne muscular dystrophy

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

[1]*Entrada Therapeutics*

432P Hydrogen sulfide attenuates skeletal muscle abnormalities in a murine model of Duchenne Muscular Dystrophy

Łoboda A[1], *Myszka M*[1,2], *Mucha O*[1], *Hajok K*[1], *Jakubczak E*[1], *Waśniowska U*[1], *Dulak J*[1]

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433P Therapeutic potential of ENTR-601-44, an Endosomal Escape Vehicle (EEV™) - Oligonucleotide Conjugate for the treatment of exon 44 skip amenable DMD

Oldham M[1], *Estrella N*[1], *Kumar A*[1], *Hicks A*[1], *Brennan C*[1], *Blake S*[1], *Li X*[1], *Pathak A*[1], *Kheirabadi M*[1], *Dougherty P*[1], *Lian W*[1], *Liu N*[1], *Gao N*[1], *Streeter M*[1], *Stadheim A*[1], *Dhanabal M*[1], *Qian Z*[1], *Girgenrath M*[1]

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434P SAT-3247: an oral small molecule inhibitor targeting AAK1, a critical effector of skeletal muscle regeneration

Mitchell R[1]

[1]*Satellos Bioscience*

435P Correction of a common Duchenne muscular dystrophy mutation by CRISPR/Cas9 gene editing using homology-independent targeted integration

Nicolau S[1,2], *Sarff J*[1], *Vetter T*[1,2], *Stephenson A*[1], *Flanigan K*[1,2]

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436P Optimizing the efficacy of antisense oligonucleotides for the treatment of Duchene muscular dystrophy

Aghaeipour A[1,2], *Torres-Masjoan L*[1], *Aguti S*[1], *Zhou H*[3,4], *Muntoni F*[1,2,4]

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437VP Cystatin C monitoring in DMD boys with viltolarsen therapy

Gremiakova T[1], *Gremiakova O*[1], *Stepanov A*[2]

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472P-496P: Genetics of NMD (new genes and NGS, diagnostic etc.)

472P Utilizing next-generation sequencing for pediatric-onset neuromuscular patients unidentified by traditional diagnostic techniques

Kulsirichawaroj P[1,2], *Sanmaneechai O*[1,2]

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473P Using RNAseq analysis in a cohort of undiagnosed congenital myopathy patients

Barraza-Flores P[1], *Genetti C*[1], *Shao W*[1], *French C*[1], *Rockowitz S*[1], *Beggs A*[1]

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474P Identifying biological pathomechanisms of TTN-affected Myopathies using RNA-sequencing data

Natraj Gayathri S[1,2], *Jonson P*[1,2], *Oghabian A*[1], *Lillback V*[1], *Sian V*[1], *Roos A*[3,4], *Hackman P*[1,2], *Udd B*[1,5], *Savarese M*[1,2]

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475P A novel clinic to resolve variants of uncertain significance in neuromuscular patients

Johnson K[1], *Schneider T*[1], *Babaian N*[1], *Azage M*[1], *McIntosh P*[1]

[1]*University Of Pennsylvania*

476P Functional studies of three novel CASQ1 variants

Laarne M[1,2], *Sarparanta J*[1,2], *Wallgren-Pettersson C*[1,2], *Jokela M*[4], *Udd B*[1,2,3], *Hackman P*[1,2], *Lehtokari V*[1,2], *Pelin K*[1,2,5]

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477P An integrated transcriptomics and genomics approach to detect an X/autosome translocation in a female with Duchenne muscular dystrophy

Segarra-Casas A[1,2], *Yépez V*[3], *Demidov G*[4], *Laurie S*[5], *Esteve A*[5], *Gagneur J*[3,6,7], *Parkhurst Y*[8], *Muni-Lofra R*[1], *Harris E*[1], *Marini-Bettolo C*[1], *Straub V*[1], *Töpf A*[1]

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478P RNA sequencing as a diagnostic tool in a cohort of 54 undiagnosed patients with neuromuscular diseases

Segarra-casas A[1], *Domínguez-González C*[2,3], *Natera-de-Benito D*[4], *Ortez C*[3,4], *Nascimiento A*[3,4], *Hernández-Laín A*[5], *Kapetanovic S*[6], *Rodríguez M*[1], *González-Mera L*[7], *Nedkova V*[7], *Fernández-Torrón R*[8], *López-de Munain A*[8], *Jimenez-Mallebrera C*[3,4], *Rodríguez-Santiago B*[1,3], *Gallardo E*[3,9], *Olivé M*[3,9], *Gallano P*[1,3], *González-Quereda L*[1,3]
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479P Hereditary Sensory & Autonomic Neuropathy Type 2A (HSAN2A): insights into genetic aetiology and variant classification

Shekhar L[1], *Lazarova A*[2], *Berry I*[2], *Majumdar A*[1]
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480P Comparative analysis of CRISPR/Cas9-targeted Nanopore long-read sequencing approaches in repeat expansion disorders

Bonne G[1,2], *Benarroch L*[1], *Boëlle P*[3], *Labrèche K*[3], *Madry H*[2], *Mohand-Oumoussa B*[2], *Eura N*[4], *Nishino I*[4], *Gourdon G*[1], *Tomé S*[1]
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481P Acute respiratory failure revealing DNAJB4 myopathy: a case report

Chitimus D[1], *Adam C*[2], *Cauquil C*[3], *Amthor S*[4], *Heming N*[4], *Annane D*[4], *Keren B*[5], *Nicolas G*[1,6], *Laforêt P*[1,6], *Metay C*[7], *Lefeuvre C*[1]
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482P Closing the gap in diagnosis of neuropathies and late-onset neurological disorders – a trans-Australia collaboration

Laing N[1,2], *Kennerson M*[3,4], *Lamont P*[5], *Vucic S*[3], *Davis M*[1,6], *Bryson-Richardson R*[7], *Ravenscroft G*[1,2], *Perez-Siles G*[3,4], *Ghaoui R*[8], *Narayanan R*[3,4], *McCombe P*[9], *Deveson I*[10], *Bryen S*[11], *Grosz B*[3,4], *Johari M*[1,2], *Rick A*[1,2], *Folland C*[1,2], *Scriba C*[1,2], *Parmar J*[1,2], *Ellis M*[3,4]
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483P Comprehensive investigation of neuromuscular manifestations in Triple-A syndrome: A combined clinical, molecular genetic and proteomic study

Oeztuerk M[1], *Nelke C*[1], *Hentschel A*[2], *Schänzer A*[3], *Meuth S*[1], *Weis J*[4], *Ruck T*[1], *Roos A*[5,6]
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484P New IRF2BPL variant in a patient with a neuromuscular phenotype

Winkler M[1], *Kornblum C*[1], *Reimann J*[1]
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485P Familial case of Bethlem myopathy caused by an ALU insertion in COL6A2

Luce L[1], Demidov G[2], Duff J[1], McFarlane A[1], Segarra Casas A[1,3], Laurie S[4,5], de Visser M[6], van der Kooi A[6], Straub V[1], Töpf A[1]

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486P Spatial analysis of skeletal muscle identifies markers of histopathological tissue alterations in Duchenne mouse models

Heezen L[1], Abdelaal T[2,3,4], van Putten M[1], Aartsma-Rus A[1], Mahfouz A[1,4,5], Spitali P[1]

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487P 2024 update of the national French consensus on gene lists for the diagnosis of muscular diseases using high-throughput sequencing

Gorokhova S[13], Pion E[1], Cossée M[2], Biancalana V[3], Acquaviva Bourdain C[4], Bouchet-Seraphin C[5], Fauré J[6], Leturcq F[7], Menassa R[8], Metay C[9], Michel-Calemard L[8], Nectoux J[7], Petit F[10], Rendu J[6], Richard P[9], Sternberg D[11], Vuillaumier-Barrot S[5], Attarian S[12], Krahn M[13]

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488P SEPN1-Related Myopathy with acute respiratory onset in middle age: a clinical and genetic study

Risi B[1], Caria F[1], Damioli S[1], Labella B[2,3], Bertella E[1], Giovanelli G[1], Ferullo L[2,3], Olivieri E[2,3], Poli L[3], Padovani A[1], Filosto M[1,2]

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489P PIEZO2-associated distal arthrogryposis: phenotypic spectrum and genotype-phenotype correlations in a multicenter case series

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490P Reclassification of missense variant pathogenicity from a hereditary spastic paraplegia gene panel using ClinGen recommendations for in silico predictor scores

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491P PIEZO2 loss of function syndrome: a highly specific and recognizable phenotype

Bharucha-Goebel D[1,2], *Lee M*[1], *Saade D*[1,3], *Donkervoort S*[1], *Yun P*[1], *Mohassel P*[1,4], *Neuhaus S*[1,5], *Ogata T*[1,6], *Averion G*[1], *Mendoza C*[1], *Alter K*[7], *Matsubara J*[7], *Stanley C*[7], *Zampieri C*[7], *Lehky T*[8], *Nickolls A*[9,10], *Chesler A*[10,11], *Foley A*[1], *Bonnemann C*[1], *and the PIEZO2 Study Team*[1]

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492P Mass spectrometry as a technique for robust quantification of titin and other large muscle disease-associated proteins

Smolnikov A[1], *Padoani D*[1], *Jurczyluk J*[1], *Su Z*[1], *Hamey J*[1], *Wilkins M*[1], *Yuen M*[1,2,3,4], *Oates E*[1,5]

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493P Gene prioritization for enhancing molecular diagnosis in rare muscle diseases of singletons

Lillback V[1], *Bergant G*[2], *Di Feo M*[1], *Bozović I*[2], *Torella A*[3], *Johari M*[1,4], *Maver A*[2], *Pelin K*[1], *Santorelli F*[5], *Nigro V*[3], *Hackman P*[1], *Peterlin B*[2], *Udd B*[1], *Savarese M*[1]

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494P Evaluation of transcriptome forward computational strategies to improve molecular diagnosis in rare pediatric neuromuscular disease

Silverstein S[1,2,3], *Ganapathy K*[4], *Donkervoort S*[1], *Moy J*[5], *Uapinyoying B*[1,6], *Gorokhova S*[7,8], *Ganesh V*[9], *Weisburd B*[9], *Orbach R*[1], *Foley A*[1], *Mohammadi P*[4,10,11,12], *Adams D*[2], *Bonnemann C*[1]

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495P Genetic mosaicism, an underestimated event in genetically unsolved neuromuscular patients: study of two families

Estévez-Arias B[1,2], *Segarra-Casas A*[3], *Ortez C*[1,4,5], *Matalonga L*[6], *Carrera-García L*[1,4], *Expósito-Escudero J*[1,4], *Jou C*[4,5,7], *Codina A*[4,7], *Jiménez-Mallebrera C*[4,5,8], *Martorell L*[5,9], *Lochmüller H*[6,10,11], *Töpf A*[12], *Beltran S*[6,8], *Hoenicka J*[2,6], *Palau F*[2,6,9,13,14], *Martí I*[15], *Gallano P*[3,5], *Nascimento A*[1,4,5], *Natera-de Benito D*[1,4], *González-Quereda L*[3,5]

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496P Recessive missense variants in DARS2 gene as novel cause of axonal Charcot-Marie-Tooth disease

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579P-595P, 596VP-597VP: EDMD, OPDM, autophagic, extramuscular

579P Myopathies associated with the FHL1 gene: a case series

Ferreira W[1], *Massaro C*[1], *Oliveira H*[1], *Badia B*[1], *Souza P*[1], *Serrano P*[1], *Farias I*[1], *Bezerra M*[1], *Pinto W*[1], *Faria Nunes K*[1], *Oliveira A*[1]

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580P SYNE-1 and SYNE-2 mutations: expanding the phenotype and genotype spectrum of Nesprin myopathy

Cheli M[1], *Marchetto G*[1], *Gibertini S*[2], *Bruno C*[3], *Filosto M*[4], *Lattanzi G*[5], *Fiorillo C*[6], *Grandis M*[7], *Malandrini A*[8], *Maioli M*[9], *Mandich P*[10], *Massa R*[11], *Matà S*[12], *Melani F*[13], *Maggi L*[2], *Rubegni A*[14], *Santorelli F*[14], *Tonin P*[1], *Cassandrini D*[14], *Vattemi G*[1]

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581P Toxic autophagic vacuolar myopathies and the role of human leukocyte antigen class I molecules and membrane attack complex in its pathogenesis

Labella B[1,2], *Lacene E*[1], *Chanut A*[1], *Leonard-Louis S*[3], *Benveniste O*[3], *Lefeuvre C*[4], *Lafôret P*[4], *Evangelista T*[1,3]

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582P Brain abnormalities in Spinal Muscular Atrophy type 1 with neurodevelopmental disorders

Holanda Mendonca R[1], *Diógenes Alencar Sindeaux R*[1], *Gontijo Camelo C*[1], *Barbante Casella E*[1], *Faria Silva Chillon K*[1], *Jose da Rocha A*[2], *Zanoteli E*[1]

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583P Recurrent pneumothoraces in chronically ventilated adult neuromuscular patients attending the National Hospital for Neurology and Neurosurgery in London

Desikan M[1], *Sir B*[1], *Astin R*[1], *Quinlivan R*[1]

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584P Nuclear envelope and splicing alterations in selected muscular dystrophies

Meinke P[1], *Todorow V*[1], *Hintze S*[1], *Schoser B*[1]

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585P Collagen XIII as the neuromuscular junction organizer

Prasannan A[1], *Norman O*[1], *Heikkinen A*[1], *Pihlajaniemi T*[1]

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586P Visualization of degenerative processes of the myofibers on muscle pathology in OPDM based on single nucleus RNA-seq data

Yamanaka A[1,2], *Eura N*[1,2], *Hayashi S*[1], *Sugie K*[2], *Noguchi S*[1], *Nishino I*[1]

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587P White matter lesions and genetic muscular diseases: an overview

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588P Phenotype variability and natural history of X-linked Myopathy with excessive autophagy running head: natural history of XMEA

Fernández-Eulate G[1], *Alfieri G[2], Spinazzi M[3], Ackermann-Bonan I[4], Duval F[5], Solé G[5], Caillon F[6], Mercier S[7], Pereon Y[8], Magot A[8], Pegat A[9], Salort-Campana E[10], Gorokhova S[11], Krahn M[11], Biancalana V[12], Evangelista T[1], Behin A[1], Metay C[13], Stojkovic T[1]*

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589P Nanopore CRISPR/Cas9-targeted sequencing reveals genetic characteristics and a possible haplotype in oculopharyngodistal myopathy

Eura N[1,2], *Noguchi S[2], Yamanaka A[1,2], Sonehara K[3], Hayashi S[2], Okada Y[3], Sugie K[1], Nishino I[2]*

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590P Genetic heterogeneity in PLIN4 gene: characterization of a new pathogenic expansion causing an autophagic vacuolar myopathy

Carnazzi A[1,2], *Iannibelli E[2], Gibertini S[2], Nicolini De Gaetano L[2], Riolo G[2], Salerno F[2], Čopič A[3], Maggi L[2], Ruggieri A[2]*

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591P DNA damage in LMNA-related congenital muscular dystrophy

Leconte M[1], *Guesmia Z[1], Bonne G[1], Bertrand A[1]*

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592P A knocked-in mouse model of GNE myopathy maintained physical function up to 70 weeks of age with supplementation of 6'-sialyl lactose

Shin J[1,3], *Choi J[1,2], Seo K[1], Kim H[1], Kim L[2], Park Y[3], Kim D[1,3]*

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593VP Emerging role of muscle and fibrosis-related microRNAs as biomarkers and therapeutic targets in Emery-Dreifuss muscular dystrophies

Bonanno S[1], *Malacarne C[1], Tarasco M[1,2], Farinazzo G[1], Mattioli E[3,4], Schena E[3,4], Saraceno F[5], Fiorillo C[6], Andreetta F[1], Schirmer E[7], Maggi L[1], Marcuzzo S[1,8], Cavalcante P[1], Lattanzi G[3,4]*

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594P *FUS/TLS* as a potential regulator of *PABPN1* in skeletal muscle?

Mouigni H[1,2,3], *Altin N[1,2,3], Ohana J[1,2,3], Kondili M[1,2,3], Dhiab J[1,2,3], Muraine L[1,2,3], Lemaitre M[1,2,3], Meunier P[1,2,3], Butler-Browne G[1,2,3], Mouly V[1,2,3], Bigot A[1,2,3], Negroni E[1,2,3], Trollet C[1,2,3]*

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595P ACTA1-related cardiomyopathy: emerging genotypic and phenotypic patterns among patients with ACTA1-related myopathy with cardiac involvement

Mcanally M[1], *Potticary A*[1], *Hayes L*[2], *Clayton J*[3], *Haliloğlu G*[4], *Donkervoort S*[1], *Yang M*[5], *Gibbons M*[6], *Gross B*[7], *Foley A*[1], *Guglieri M*[8], *Lee T*[9], *Munot P*[10], *Sarkozy A*[10], *Muntoni F*[10,11], *Straub V*[8], *Laing N*[3,12], *Beggs A*[13], *Bönnemann C*[1], *for the ACTA1-Cardiomyopathy Study Group*[14,15,16,17,18,19,20,21,22]

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596VP Elevated VCP ATPase activity correlates with disease onset in multisystem proteinopathy-1

Robinson S[1], *Findlay A*[1], *Schiava M*[2], *Daw J*[1], *Diaz-Manera J*[2], *Chou T*[3]

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597VP Vacuolar myopathy with permanent weakness: case report with imaging, muscle biopsy, and molecular features

Cotta A[1], *Carvalho E*[1], *da-Cunha-Júnior A*[1], *da Silveira E*[2], *Costa-e-Silva C*[2], *Nunes-Neves S*[1], *Valicek J*[1], *Vargas A*[1]

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619P-642P, 643VP: FSHD

619P Normal and borderline-sized D4Z4 alleles in FSHD1-mimics: a multicentric Italian review of cases

Gadaleta G[1], *Vercelli L*[1], *Urbano G*[1], *Rolle E*[1], *Torri F*[2], *Pugliese A*[3], *Maggi L*[4], *Tupler R*[5], *Filosto M*[6], *Rodolico C*[3], *Ruggiero L*[7], *Ricci G*[2], *Siciliano G*[2], *Mongini T*[1]

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620P Novel outcome measures to quantify longitudinal changes in motor function in FSHD

Hayward L[1], *Andrade E*[1], *Ridout B*[2], *Penka J*[2], *Stauffer J*[2], *Tulchin-Francis K*[3], *Lowes L*[3], *King O*[1], *Watts G*[1]

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621P The rarest subtype of a rare disease: Infantile Facio-Scapulo-Humeral Dystrophy

Yunisova G[1], *Arduç Akçay A*[2], *Eraslan S*[3], *Avci Ş*[3], *Eren İ*[4], *Özdağ Acarli A*[5], *Kayserili H*[3], *Oflazer P*[5]

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622P An unusual case of double trouble of Facioscapulohumeral dystrophy (FSHD) and coexisting congenital myopathy uncovered by whole genome sequencing

Kulshrestha R[1], *Emery N*[1], *Strachan K*[1], *Willis T*[1,2], *Sewry C*[1], *Morley-Davies A*[3]

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623P Muscular Dystrophy in the Japanese nationwide registry of muscular dystrophy (Remudy)

Takizawa H[1], *Yoshioka W*[2], *Mori-Yoshimura M*[1], *Saito Y*[2], *Nishino I*[2], *Nakamura H*[3], *Matsumura T*[4]

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624P A new double-trouble phenotype: fascioscapulohumeral muscular dystrophy and familial amyotrophic lateral sclerosis

Alonso-pérez J[1], *León-Hernández J*[1], *Henao-Ramírez M*[1], *Méndez-Hernández L*[2], *Sánchez R*[2], *Martínez-Bugallo F*[3], *Prieto-Morín C*[3]

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625P From phenotype to genotype: diagnosis pitfalls in atypical FSHD cases

Torri F[1], *Strafella C*[2], *Vercelli L*[3], *Gadaleta G*[3], *Risi B*[4], *Colantoni L*[2], *Giardina E*[2], *Filosto M*[4], *Mongini T*[3], *Siciliano G*[1], *Ricci G*[1]

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626P Disability questionnaire of FSHD1 correlates with the in-person examination

Lee J[1], *Shin J*[2], *Shin J*[3]

[1]*Kyungpook National University Hospital,* [2]*Pusan National University Yangsan Hospital, Yangsan,* [3]*Hallym medical center, Dongtan Sacred Heart Hospital*

627P The FORCE(TM) platform achieves robust and durable DUX4 suppression and improves muscle function in Facioscapulohumeral muscular dystrophy mouse model

Picariello T[1], *Natoli T*[1], *Yoder N*[1], *Yao M*[1], *Valdivia B*[1], *Tahaei E*[1], *Johnson J*[1], *Qiu Q*[1], *More P*[1], *Schlaefke L*[1], *Yang S*[1], *Basiri B*[1], *Weeden T*[1], *Beskrovnaya O*[1], *Zanotti S*[1]

[1]*Dyne Therapeutics*

628P Development of a CRISPR/CasX 4q telomeric region ablation strategy for FSHD1 using an isogenic hiPSC line and a FSHD1 fibroblast cell line

Lama C[1], *de Graaf N*[1,2], *Bou Akar R*[1], *Danaus P*[3], *Suel-Petat L*[4], *Nectoux J*[3], *Authier F*[1,5], *Relaix F*[1], *Richard I*[4], *Malfatti E*[1,5]

[1]*Université Paris Est Créteil, INSERM, IMRB - Hôpital Henri-Mondor,* [2]*The Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud university medical center,* [3]*Assistance Publique - Hôpitaux de Paris, APHP, Centre Universitaire Paris, Hôpital Cochin, Laboratoire de Génétique et Biologie Moléculaires,* [4]*Généthon, INSERM, Unité de recherche Integrare, UMR_S951 - Université Paris-Saclay, Université d'Évry,* [5]*Reference Center for Neuromuscular Disorders, APHP Henri Mondor University Hospital*

629P Co-producing UK care quality standards in Facioscapulohumeral muscular dystrophy (FSHD) in partnership with people with FSHD, carers and healthcare professionals: a qualitative focus group study

Leone E[1], *Pandyan A*[2], *Rogers A*[1], *Kulshrestha R*[3], *Hill J*[1], *Philp F*[4]

[1]*Keele University,* [2]*Bournemouth University,* [3]*The Robert Jones and Agnes Hunt Orthopaedic Hospital, Gobowen,* [4]*University of Liverpool*

630P Genetic diversity and clinical implications of Facioscapulohumeral muscular dystrophy in the Indian population

Lemmers R[1], *Venugopalan Y V*[2], *van der Vliet P*[1], *Reyaz A*[2], *Mishra R*[2], *Ahmad T*[2], *Kretkiewicz M*[1], *Macken W*[3], *Efthymiou S*[3], *Bhatia R*[2], *Dominik N*[3], *Houlden H*[3], *Morrow J*[3], *Wilson L*[3], *Hanna M*[3], *Bugiardini E*[3], *van der Maarel S*[1], *Padma Srivastava M*[2]

[1]*Leiden University Medical Centre,* [2]*All India Institute of Medical Sciences,* [3]*University College London*

631P Respiratory function and trajectories in Facioscapulohumeral muscular dystrophy (FSHD): a clinical audit

Robinson E[1], *Lees E*[1], *Moat D*[1], *Michell-Sodhi J*[1], *Richardson M*[1], *Wong K*[1], *Grover E*[1], *Schiava M*[1], *Bolano C*[1], *Salman D*[1], *Guglieri M*[1], *Elseed M*[1], *McCallum M*[1], *Diaz-Manera J*[1], *James M*[1], *Straub V*[1], *Marini Bettolo C*[1], *Pace F*[1], *Muni-Lofra R*[1], *Tasca G*[1]

[1]*John Walton Muscular Dystrophy Research Centre*

632P A human skeletal muscle-on-chip model for Facioscapulohumeral muscular dystrophy: improving maturation and complexity

Augustinus R[1], *Franken M*[1], *van der Maarel S*[1], *Pijnappel P*[2], *de Greef J*[1]

[1]*Leiden University Medical Center,* [2]*Erasmus Medical Center*

633P Penetrance of 5RU founder allele and correlation of D4Z4 methylation levels to severity in large kindred with FSHD

Butterfield R[1], *Dunn D*[2], *Duval B*[2], *Moldt S*[1], *Weiss R*[2]

[1]*Unviersity of Utah, Department of Pediatrics,* [2]*Unviersity of Utah, Department of Human Genetics*

634P Retrospective analysis of muscle biopsy findings in a cohort of patients with Facioscapulohumeral Dystrophy Type 1

Ruggiero L[1], *Pezzella M*[1], *Di Leo R*[1], *Vitale F*[1], *Russo R*[1], *Grasso C*[1], *Iapoce M*[1], *Boemia V*[1], *Tupler R*[2], *Fiorillo C*[3], *Zoppi D*[1]

[1]*Policlin Universita Federico Ii - Clinica 2 Neurologia,* [2]*Department of Biomedical, Metabolic and Neural Sciences, University of Modena and Reggio Emilia, Modena.,* [3]*Department of Neurosciences, Rehabilitation, Ophthalmology, Genetic and Maternal and Infantile Sciences, University of Genoa,*

635P Pain impacts quality of life, psychological disorders and exercise in a large international cohort of patients with facioscapulohumeral muscular dystrophy

Knox R[1], *Wang L*[2], *Elsheikh B*[3], *LoRusso S*[4], *Zhao S*[3], *Eichinger K*[5], *Higgs K*[6], *Lewis L*[5], *Walker M*[6], *Sansone V*[7], *Leung D*[8], *Sacconi S*[9], *Mul K*[10], *Shieh P*[11], *Butterfield R*[12], *Johnson N*[13], *Bugiardini E*[14], *McDermott M*[5], *Tawil R*[5], *Statland J*[6]

[1]*Washington University School Of Medicine,* [2]*University of Washington,* [3]*The Ohio State University,* [4]*Kaiser Permanente,* [5]*University of Rochester Medical Center,* [6]*University of Kansas Medical Center,* [7]*The NEMO Clinical Center, University of Milan,* [8]*Kennedy Krieger Institute,* [9]*Nice University,* [10]*Radboud University,* [11]*University of California,* [12]*University of Utah,* [13]*Virginia Commonwealth University,* [14]*University College London*

636P Longitudinal insights into childhood onset Facioscapulohumeral dystrophy: a five-year natural history study

Dijkstra J[1,2], *Boon H*[1,2], *Koekkoek A*[2], *Goselink R*[3], *Pelsma M*[4], *Houwen-van Opstal S*[4], *Van Alfen N*[5], *Van Engelen B*[1], *Voermans N*[1], *Erasmus C*[2]

[1]*Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center,* [2]*Department of Pediatric Neurology, Donders Institute for Brain, Cognition and Behaviour, Amalia Children's Hospital, Radboud University Medical Centre,* [3]*Department of Neurology, Jönköping, Region Jönköping County, and Department of Biomedical and Clinical Sciences, Linköping University,* [4]*Department of Rehabilitation, Donders Institute for Brain, Cognition and Behaviour, Amalia Children's Hospital, Radboud University Medical Center,* [5]*Department of Neurology, Clinical Neuromuscular Imaging Group, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center*

637P RelnForce: A bicentric, randomized, double blind, placebo-controlled pilot study to evaluate the efficacy and safety of satralizumab in FSHD1

Pini J[1,2], *Aleman A*[3], *Breiner A*[3], *Cavalli M*[1], *Puma A*[1], *Villa L*[1], *Gaki E*[4], *McIver T*[4], *Okumu S*[5], *Sidiropoulos P*[5], *Lochmüller H*[3], ***Saconni S***[1,2]

[1]*Peripheral Nervous System and Muscle Department, University Côte d'Azur, CHU Nice,* [2]*University Cote d'Azur, Inserm, CNRS, Institute for Research on Cancer and Aging of Nice (IRCAN),* [3]*Ottawa Hospital Research Institute,* [4]*Roche Products Ltd, 5F. Hoffmann-La Roche Ltd*

638P Interleukin-6 as a Biomarker for Disease Activity, Progression and Muscle Composition in Facioscapulohumeral Dystrophy: Insights from Longitudinal Studies

Saconni S[1,2], *PINI J*[1,2], *Martinuzzi E*[3], *Puma A*[1], *Villa L*[1], *Cavali M*[1], *Ezaru A*[1], *Garcia J*[1], *Glaichenhaus N*[3]

[1]*Peripheral Nervous System and Muscle Department, University Côte d'Azur, CHU Nice,* [2]*University Cote d'Azur, Inserm, CNRS, Institute for Research on Cancer and Aging of Nice (IRCAN),* [3]*University Cote d'Azur, CNRS, Inserm, Institute of Molecular and Cellular Pharmacology, Sophia Anitpolis, France*

639P **The other face of Facioscapulohumeral Muscular Dystrophy: exploring facial weakness using muscle ultrasound**

Vincenten S[1], *Van Doorn J*[1], *Teeselink S*[1], *Rasing N*[1], *Padberg G*[1], *Voermans N*[1], *van Engelen B*[1], *van Alfen N*[1], *Mul K*[1]

[1]*Radboudumc, department of Neurology*

640P Efficacy and safety of EPI-321, an investigational single dose Epigenome editing therapy targeting D4Z4 in Facioscapulohumeral Muscular Dystrophy (FSHD)

Boregowda S[1], *Zheng H*[1], *Agrawal V*[1], *Norton A*[1], *Wasala N*[1], *Collin de l'Hortet A*[1]
[1]*EPICRISPR Biotechnologies, Inc.*

641P Modeling cell type specific and sporadic *DUX4* gene expression in FSHD

Sasaki-Honda M[1,2], *Sakurai H*[1], *Rada-Iglesias A*[2]
[1]*CiRA, Kyoto University,* [2]*IBBTEC, University of Cantabria*

642P 4qA D4Z4 methylation test as a valuable complement for differential diagnosis in patients with FSHD-like phenotype

Xia X[1], *Cheng N*[1], *Liu Y*[1], *Zhu W*[1], *Zhao C*[1]
[1]*Huashan Hospital, Fudan University*

643VP Molecular imaging of muscle involvement in Facioscapulohumeral Muscular Dystrophy using multispectral optoacoustic tomography

Monforte M[1], *Bortolani S*[1], *Ravera B*[2], *Marchese D*[2], *Qiu Y*[3], *Burton N*[3], *Torchia E*[2], *De Spirito M*[2], *Ricci E*[2], *Tasca G*[4]
[1]*Fondazione Policlinico Universitario A. Gemelli IRCCS,* [2]*Università Cattolica del Sacro Cuore,* [3]*iThera Medical GmbH,* [4]*John Walton Muscular Dystrophy Research Centre, Newcastle University*

677P-686P, 687P: RNA in NMD: clinical insights, pathomechanisms and treatments

677P The Splice Index as a prognostic biomarker of strength and function in Myotonic Dystrophy type 1

Provenzano M[1,2], *Ikegami K*[1,2], *Bates K*[1,2], *Hartman J*[1,2], *Jones A*[1,2,3], *Butler A*[1,2,3], *Berggren K*[1,2], *Hung M*[4], *Kiefer M*[1,2,5], *Thornton C*[6,7], *Johnson N*[1,2], ***Hale M***[1,2], *on behalf of the Myotonic Dystrophy Clinical Research Network (DMCRN)*
[1]*Department of Neurology, Virginia Commonwealth University,* [2]*Center for Inherited Muscle Research, Virginia Commonwealth University,* [3]*Children's Hospital of Richmond at Virginia Commonwealth University, Pediatric Therapy Services,* [4]*College of Dental Medicine, Roseman University of Health Sciences,* [5]*Department of Physical Therapy, Virginia Commonwealth University,* [6]*Department of Neurology, University of Rochester School of Medicine and Dentistry,* [7]*Center for RNA Biology, University of Rochester School of Medicine and Dentistry*

678P Unraveling calcium dysregulation and autoimmunity in immune mediated Rippling muscle disease

Nath S[1], *Dasgupta A*[2], *Dubey D*[1], *Kokesh E*[1], *Liewluck T*[1], *Beecher G*[3], *Pittock S*[1], *Doles J*[2], *Litchy W*[1], *Milone M*[1]
[1]*Mayo Clinic Department of Neurology,* [2]*Department of Anatomy, Cell Biology, & Physiology, Indiana University,* [3]*Faculty of Medicine & Dentistry - Medicine Dept, University of Alberta*

679P Clinical and molecular characterization of DMD pseudo-3'-terminal exons

Frair E[1], *Nicolau S*[1,2,3], *Meyer* [A1,2,4], *Vetter T*[1,2], *Dunn D*[5], *Gabel L*[1], *Waldrop M*[1,2,3], *Weiss R*[5], *Flanigan K*[1,2,3]
[1]*Center for Gene Therapy, Abigail Wexner Research Institute, Nationwide Children's Hospital,* [2]*Department of Pediatrics, The Ohio State University College of Medicine,* [3]*Department of Neurology, The Ohio State University,* [4]*Division of Genetic and Genomic Medicine, Nationwide Children's Hospital, 5Department of Human Genetics, The University of Utah*

680P Developing effective RNA editing approaches to target the nonsense mutation in the key functional region of *DMD*

Lam H[1,2], *Uzwyshyn-Jones K*[1], *Ma S*[1], *Muntoni F*[2], *Meng J*[1,3], *Zhou H*[1,3]
[1]*Genetic and Genomic Medicine Department, Great Ormond Street Institute of Child Health, University College London,* [2]*The Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health,* [3]*NIHR Great Ormond Street Hospital Biomedical Research Centre, University College London*

681P A novel COL11A1 variant in a child with neuromuscular findings: expanding the genotypic and phenotypic spectrum of COL11A1-related disease

Mcanally M[1], *Potticary A*[1], *Donkervoort S*[1], *Hu Y*[1], *Huryn L*[3], *Pais L*[2], *Harper A*[4], *Foley A*[1], *Bönnemann C*[1]
[1]*Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, NIH,* [2]*Program in Medical and Population Genetics, Broad Institute of MIT and Harvard,* [3]*National Eye Institute, NIH,* [4]*Department of Neurology, Division of Child Neurology, Children's Hospital of Richmond at VCU*

continued on next page

682P Differential splicing of *OBSCN* throughout human cardiac and skeletal muscle development

Oghabian A[1,2], *Jonson P*[1,3], *Johari M*[1,12], *Andres D*[4], *Munell F*[5], *Soriano J*[6], *Duran M*[7], *Sinisalo J*[8], *Tolppanen H*[9], *Tolva J*[10], *Hackman P*[1,3], *Savarese M*[1,3], *Udd B*[1,3,4,11]

[1]*Folkhälsan Research Center,* [2]*Research Program for Clinical and Molecular Metabolism, Faculty of Medicine, University of Helsinki,* [3]*Department of Medical Genetics, Medicum,* [4]*Department of Neurology, Vaasa Central Hospital,* [5]*Child Neurology Unit. Hospital Universitari Vall d'Hebron, Vall d'Hebron Research Institute (VHIR),* [6]*Unitat De Malalties Neuromusculars Pediàtriques, Hospital Universitari Vall D'hebron,* [7]*Histology Department, Vall D'hebron University Hospital,* [8]*Maternal Fetal Medicine Unit, Department of Obstetrics, Universitat Autònoma de Barcelona, Hospital Vall D'hebron,* [9]*Helsinki University Central Hospital,* [10]*Transplantation Laboratory, Department of Pathology,* [11]*Neuromuscular Research Center, Department of Neurology, Tampere University and University Hospital,* [12]*Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia*

683P Phase IIa trial assessing the efficacy and safety of Enpatoran in patients with Dermatomyositis and Polymyositis: NEPTUNIA rationale and study design

Aggarwal R[1], *Roy S*[2], *Thömmes G*[3], *Weinelt D*[3], *Chitkara D*[4], ***Denis D***[5]

[1]*University of Pittsburgh,* [2]*Ares Trading SA, an affiliate of Merck KGaA,* [3]*Merck Healthcare KGaA,* [4]*EMD Serono Research & Development Institute, Inc., an affiliate of Merck KGaA,* [5]*EMD Serono, Inc., an affiliate of Merck KGaA*

684P Complex duplications containing terminal exon duplications in the DMD gene

Meyer A[1,2], *Nicolau S*[1], *Dunn D*[3], *Gabel L*[1], *Frair E*[1], *Weiss R*[3], *Flanigan K*[1,2]

[1]*Nationwide Children's Hospital,* [2]*The Ohio State University,* [3]*University of Utah*

685VP Reduction of dystrophin-targeting microRNAs increases expression of clinically relevant dystrophin isoforms

Fiorillo A[1,2], *McCormack N*[2], *Calabrese K*[2], *Sun C*[2], *Tully C*[2], *Heier C*[1,2]

[1]*Virginia Commonwealth University,* [2]*Children's National Hospital*

686P Identification and characterization of actionable deep intron 8 *IGHMBP2* hotspot pathogenic variants through motor neuron RNAseq and whole genome

Silverstein S[1,2,3], *Donkervoort S*[1], *Cassini T*[4], *Vetrini F*[5,6], *Conboy E*[5,6], *Comer A*[7], *Treat K*[5,6], *Liaqat K*[5,6], *Mantcheva L*[5,6], *Patankar A*[8], *Bharucha-Goebel D*[1,9], *Foley A*[1], *Chao K*[10], *Neuhaus S*[1], *Grunseich C*[8], *Bonnemann C*[1]

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687P Splicing switching of alternative last exons due to a deletion including canonical polyadenylation site in *COL6A2* gene causes recessive UCMD

Saito Y[1], *El Sherif R*[2,3], *Awaya T*[4], *Hayashi S*[1], *Noguchi S*[1], *Nishino I*[1]

[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry,* [2]*Myo-Care Neuromuscular Center, Myo-Care National Foundation,* [3]*School of Medicine, New Giza University,* [4]*Department of Anatomy and Developmental Biology, Graduate School of Medicine and Faculty of Medicine, The University of Kyoto*

18:15-18:45	**Short Oral Presentations 4** North Hall **641P, 630P, 638P, 639P, 640P** *Moderator: Nicol Voermans, Radboud University Medical Centre*	**Short Oral Presentations 5** Terrace 2A **586P, 50P, 97P, 96P, 51P** *Moderator: Piera Smeriglio. Institut de Myologie*	**Short Oral Presentations 6** Terrace 2B **496P, 686P, 594P, 595P, 403P** *Moderator: Nigel Laing, Harry Perkins Institute of Medical Research*
19:00-20:00	**Industry Symposium 5** South Hall 1 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium	**Industry Symposium 6** South Hall 2 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium	

Thursday 10th October 2024

07:00-15:00	**Congress desk open**	
07:15-08:15	**NMD Board Meeting** Club E (separate registration required)	
08:15-09:15	**Industry Symposium 7** South Hall 1 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium	**Industry Symposium 8** South Hall 2 **Catering provided by the sponsor for this session**, available from 30 minutes prior to the start of the symposium
09:30-11:00	**Topic 2: NMD Around the World** Congress Hall *Moderators: Hernan Gonorazky, The Hospital for Sick Children, Canada & Rasha El Sherif, Newgiza University, Egypt* **06INV** What about management of neuromuscular diseases in Latin America? ***Monges S***[1] [1]*Hospital de Pediatría J.P. Garrahan* **07INV** Neuromuscular disorders in India: New World meets the Old ***Venugopalan Y V***[1] [1]*All India Institute of Medical Sciences* **05INV** Advancing neuromuscular disorders in Senegal, West Africa, through collaborative networks and available resources ***Rodriguez Cruz P***[1,2,3], [1]*Centro Nacional de Análisis Genómico (CNAG),* [2]*Neurology Department, CHNU de Fann,* [3]*Université Cheikh Anta Diop de Dakar* **08INV** An African perspective on muscle diseases ***Heckmann J***[1] [1]*University of Cape Town*	
11:00-11:30	**Morning refreshments, exhibition and posters** Congress Hall Foyer, Forum Hall and Forum Foyer	
11:00-11:30	**Myology Café - Meet the WMS Myology Developments Across the World Committee** Congress Hall Foyer	
11:30-13:30	**Topic 2: NMD Around the World** Congress Hall *Moderators: Sharon Aharoni, Schneider Childrens' Medical Center, Israel & Jorge Bevilacqua, Hospital Clínico Universidad De Chile & Clinica Dávila, Chile*	
11:30-11:45	**07O** Rare neuromuscular disease specific mega clinics: a proposal to bridge the gap in resource limited settings ***Ramesh Babu R***[1,2], *Krishna G*[1,2], *Satyam P*[1,2], *Rao S*[3], *Maganthi M*[2], *Agnes Mathew A*[1,2] [1]*Synapse Neuro Centre & Child Development Centre, Bangalore Baptist Hospital,* [2]*Bangalore Baptist Hospital,* [3]*Organization of Rare Diseases*	
11:45-12:00	**08O** To manually or mechanically cough: that is the question. Cough augmentation in children with neuromuscular disorders: a feasibility study ***Human A***[5], *Corten L*[3], *Morrow B*[4] [1]*Department of Physiotherapy, School of Health Care Sciences, Sefako Makgatho Health Sciences University,* [2]*Department of Health and Rehabilitation Sciences (Division Physiotherapy),* [3]*School of Sport and Health Sciences (Physiotherapy), University of Brighton,* [4]*Department of Paediatrics and Child Health, University of Cape Town,* [5]*Department of Physiotherapy, School of Health Care Sciences, University of Pretoria*	
12:00-12:15	**09O** The clinical and molecular landscape of genetic neuromuscular disorders in Senegal, West Africa ***Rodriguez Cruz P***[1,2,3], *Diagne R*[2,3], *Diop A*[4], *Diouf A*[5], *Ndong M*[6], *Senghor H*[2], *Dieng S*[6], *Oko C*[2], *Traoré M*[7], *Mbaye K*[8], *Seck L*[2], *Sy M*[2], *Sow A*[2,9], *Diagne N*[2], *Martorell Sampol L*[10], *Nascimiento A*[10], *Beltran S*[1], *Ndiaye R*[3], *Ndiaye M*[2,3], *Diop A*[2,3] [1]*Centro Nacional de Análisis Genómico (CNAG),* [2]*Neurology Department, CHNU de Fann,* [3]*Université Cheikh Anta Diop de Dakar,* [4]*Neurology Department, Centre Hospitalier National de Pikine,* [5]*Centre Talibou Dabo,* [6]*Centre Hospitalier National d'Enfants Albert Royer,* [7]*Centre Hospitalier Roi Baudouin,* [8]*Centre Hospitalier Régional de Ziguinchor,* [9]*Centre Hospitalier National pour Enfants de Diamniadio,* [10]*Hospital San Joan de Deu*	

12:15-12:30	**10O** The neonatal screening of SMA in Ukraine: the 18 months of experience ***Olkhovych N***[6], *Servais L*[2], *Makukh H*[3], *Grechanina O*[4], *Veropotvelyan M*[5], *Samonenko N*[6], *Mytsyk N*[2], *Barvinskaya O*[2], *Kormoz S*[2], *Shklyarskaya T*[6], *Gorovenko N*[7] *[1]Expert Centre of Neonatal Screening, [2]MDUK Oxford and NIHR, University of Oxford, [3]Lviv Regional Centre of Neonatal Screening, [4]Kharkiv Regional Centre of Neonatal Screening, [5]Kryviy Rig Regional Centre of Neonatal Screening, [6]National Children's Specialized Hospital "OHMATDYT" MoH, [7]National University of Health Care named after P.L. Shupika*
12:30-12:45	**11O** Genetic profile of Brazilian patients with LAMA2-related dystrophies ***Camelo C***[1], *Moreno C*[1], *Artilheiro M*[1], *Fonseca A*[1], *Gianetti J*[2], *Barbosa A*[3], *Donis K*[4], *Saute J*[4], *Pessoa A*[5], *VanderLinden H*[6], *Gonçalves A*[7], *Kulikowski L*[1], *Kok F*[1], *Zanoteli E*[1] *[1]University Of Sao Paulo, [2]Universidade Federal de Minas Gerais, [3]Fundação Hospitalar do Estado de Minas Gerais, [4]Hospital de Clínicas de Porto Alegre, [5]Children's Hospital Albert Sabin, [6]Rehabilitation Center Dr. Henrique Santillo, [7]University of Porto*
12:45-13:00	**12O** Clinical, paraclinical features and classification of myositis in Vietnam ***Le S***[1], *Nguyen Le Trung H*[2] *[1]University Medical Center of Hochiminh, [2]University of Medicine and Pharmacy of Hochiminh*
13:00-13:15	**13O** Genomic neuromuscular disorders in Turkey originate from a vast background ***Topaloğlu H***[1] *[1]Yeditepe University, Pediatrics*
13:15-13:30	**14O** Inherited Neuromuscular disorders in India: Outcomes of 1000 probands in the ICGNMD study at AIIMS New Delhi ***Venugopalan Y V***[1], *Macken W*[2,3], *Wilson L*[2], *Rani N*[1], *Reyaz A*[1], *Ahmad T*[1], *Dalal A*[4], *Vandrovcova J*[2], *Dominik N*[2], *Consortium ICGNMD-Consortium*[5], *Tallapaka K*[6], *Lemmers R*[7], *Reilly M*[2], *Hanna M*[2,3], *Bhatia R*[1], *Pitceathly R*[2,3], *Houlden H*[2], *Thangaraj K*[4,6], *Straub V*[8], *Srivastava P*[1] *[1]All India Institute of Medical Sciences, [2]Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, [3]NHS Highly Specialised Service for Rare Mitochondrial Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery, [4]Centre for DNA Fingerprinting and Diagnostics, Hyderabad, [5]https://www.ucl.ac.uk/genomic-medicine-neuromuscular-diseases/global-contributor-list, [6]Centre for Cellular & Molecular Biology, [7]Department of Human Genetics, Leiden University Medical Center (LUMC), [8]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust*
13:30-14:45	**Lunch, exhibition and posters** Congress Hall Foyer, Forum Hall and Forum Foyer
13:30-14:45	**Career Development Session - Engagement with industry** Zoom and Panorama Rooms, 1st floor (separate registration required and lunch is provided)
15:00-17:30	**Poster viewing / Group activity** (separate registration required) Please refer to your joining instructions for further details of group activities and schedules.
17:30-20:00	**Group activity networking reception** Kaiserstein Palace (separate registration required)

Friday 11th October 2024

06:45-17:15	**Congress desk open**
07:00	**Light refreshments available**
07:30-08:30	**Interesting Case Discussions** 📍 South Hall 2 *Moderators: A. Reghan Foley, National Institutes of Health, USA & Hans-Hilmar Goebel, Charité – Universitätsmedizin Berlin, Germany* **Case 1:** HIV-associated inclusion body myositis in a young woman *Jeannine Heckmann, University of Cape Town, South Africa* **Case 2:** A muscle disorder presenting with Resistant Hypercalcaemia *Ashirwad Merve, University College London Hospitals, UK* **Case 3**: TUBA4A associated congenital myopathy with centronuclear findings and protein aggregates *Cristiane Moreno, Universidade de São Paulo, Brazil* **Case 4**: Proximal and facial weakness: Beyond FSHD *Carmen Paradas Lopez, Virgen del Rocío Hospital, Spain* **Case 5**: ADSSL1 -associated myopathy presented with EDMD like phenotype *Hacer Durmus, Istanbul Faculty of Medicine, Turkey*
08:30-08:45	**Comfort break**
08:45-10:15	**Topic 3: RNA in NMD: Clinical Insights, Pathomechanisms and Treatments** 📍 Congress Hall *Moderators: Liubov Gushchina, The Abigail Wexner Research Institute, Nationwide Children's Hospital, USA & Emma Rybalka, Victoria University, Australia*
08:45-09:15	**09INV** An overview of the RNA pathomechanisms in neuromuscular diseases, ***Swanson M***[1] [1]*University of Florida College of Medicine*
09:15-09:45	**10INV** Clinical and diagnostic utility of RNA/transcriptome: An overview of role of RNA in disease gene discovery and diagnostics ***Yoon G***[1] [1]*The Hospital for Sick Children*
09:45-10:00	**15O** Spatial transcriptomics analysis of Becker and Duchenne skeletal muscle to decipher histopathological alterations in dystrophinopathies ***Heezen L***[1], *Mao Q*[1], *van Putten M*[1], *Diaz Manera J*[2], *Kan H*[3], *Niks E*[4], *Aartsma-Rus A*[1], *Nicolau S*[5], *Flanigan K*[5,6,7], *Mahfouz A*[1,8,9], *Spitali P*[1] [1]*Department of Human Genetics, Leiden University Medical Center,* [2]*Faculty of Medical Sciences, Newcastle University,* [3]*Department of Radiology, Leiden University Medical Center,* [4]*Department of Neurology, Leiden University Medical Center,* [5]*Center for Gene Therapy, The Abigail Wexner Research Institute at Nationwide Children's Hospital,* [6]*Department of Pediatrics, The Ohio State University,* [7]*Department of Neurology, The Ohio State University,* [8]*Delft Bioinformatics Lab, Delft University of Technology,* [9]*Leiden Computational Biology Center, Leiden University Medical Center*
10:00-10:15	**16O** Lost in translation: pathogenic translation of GGC repeats in novel and toxic proteins in Oculopharyngodistal myopathy (OPDM) ***Boivin M***[1], *Schmitt L*[1], *Grandgirard E*[1], *Morlet B*[1], *Negroni L*[1], *Goetz-Reiner P*[1], *Lefebvre E*[1], *Maglott A*[1], *Eberling P*[1], *Oulad-Abdelghami M*[1], *Deng J*[2], *Charlet-Berguerand N*[1] [1]*IGBMC,* [2]*Peking University First Hospital*
10:15-10:45	**Morning refreshments, exhibition and posters** 📍 Congress Hall Foyer, Forum Hall and Forum Foyer
10:15-10:45	**Myology Café - Meet the WMS Sustainability Committee** 📍 Forum Hall Foyer
10:45-12:15	**Topic 3: RNA in NMD: Clinical Insights, Pathomechanisms and Treatments** 📍 Congress Hall *Moderators: Gisèle Bonne, Institut de Myologie, France & Silvère Van Der Maarel, Leiden University Medical Center, The Netherlands*
10:45-11:15	**11INV** RNA as biomarkers in neuromuscular diseases, including monitoring progression, in clinical trial design, as outcomes ***Spitali P***[1] [1]*Leiden University Medical Center*
11:15-11:45	**12INV** RNA therapeutics in neuromuscular diseases, including new developments, and challenges ***Arechavala Gomeza V1***[1,2] [1]*Biobizkaia Health Research Institute,* [2]*Ikerbasque, Basque Foundation for Science*

11:45-12:00	**17O** Schwann cell transduction and PMP22 target engagement in non-human primates supports translation of RNAi-based gene therapy for CMT1A ***Wallace L***[1], *Thangaraj M*[1], *Stavrou M*[2], *Price B*[3], *Zender G*[1], *Bayazit M*[1], *Taylor N*[1], *Kleopa K*[2], *Harper S*[1,4] [1]*Nationwide Children's Hospital,* [2]*The Cyprus Institute of Neurology & Genetics,* [3]*Armatus Bio,* [4]*Department of Pediatrics, The Ohio State University College of Medicine*
12:00-12:15	**18O** Breaking ground in CMT1B treatment: AAV9-mediated dual RNAi and gene replacement therapy targeting schwann cells improves myelination and peripheral nerve function in mice ***McCulloch M***[1,2], *Munezero D*[1], *Paripati A*[1], *Zhu J*[1], *Amini Chermahini G*[1], *Chuah R*[1], *Rashnonejad A*[1,2,3] [1]*Nationwide Children's Hospital (Center for Gene Therapy),* [2]*The Ohio State University (Molecular, Cellular, and Developmental Biology Graduate Program),* [3]*The Ohio State University (Department of Pediatrics)*
12:30-13:30	**WMS General Assembly / Poster viewing for non-members** South Hall 2
12:30-14:00	**Lunch, exhibition and posters** Congress Hall Foyer, Forum Hall and Forum Foyer
13:45-14:15	**Sponsor Meeting** South Boardroom 1
14:15-15:15	**Poster Session 3** Forum Hall (refreshments provided)

55P-75P, 76VP-78VP: Distal myopathies, MFM

55P Novel mutations and genotype-phenotype correlation in a multicenter cohort of GNE myopathy in China
Jiao K[1], *Zhang J*[1], *Li Q*[2], *Lv X*[3], *Hong D*[6], *Zhao Z*[4], *Wang Z*[5], *Zhu W*[1]
[1]*Department of Neurology and Rare Disease Center, Huashan Hospital, Fudan University, and National Center for Neurological Disorders (NCND),* [2]*Xiangya Hospital, Central South University,* [3]*Department of Neurology and Research Institute of Neuromuscular and Neurodegenerative Diseases, Qilu Hospital of Shandong University,* [4]*Department of Neuromuscular Disease, Third Hospital of Hebei Medical University,* [5]*The First Affiliated Hospital of Fujian Medical University,* [6]*Dtment of Neurology and Department of Medical Genetics, The First Affiliated Hospital of Nanchang University*

56P Mutational screening of Distal Myopathy gene *ACTN2* reveals an aggregation hot-spot in the actin-binding domain
Ranta-aho J[1,2], *Jonson P*[1,2], *Sarparanta J*[1,2], *Udd B*[1,3], *Savarese M*[1,2]
[1]*Folkhälsan Research Center,* [2]*University of Helsinki,* [3]*Tampere University and Tampere University Hospital*

57P Phenotypic description of 40 patients with p.Ser55Phe variant in the *MYOT* gene: MYOT-MUR study
Martínez Marín R[1], *Aledo Serrano M*[1], *Mena Bravo A*[1], *Lorenzo Diéguez M*[1], *García Leal A*[1], *Zmork Martínez G*[1], *Pérez Lucas J*[2], *Pérez García E*[1]
[1]*Hospital Universitario La Paz,* [2]*Hospital Universitario Del Tajo*

58P *ADSSL1* myopathy in a neuromuscular cohort in Northern India
Reyaz A[1], *Macken W*[2,3], *Rani N*[1], *Ahmad T*[1], *Tarane K*[1], *Danish M*[1], *ICGNMD Consortium*[5], *Bhatia R*[1], *Pitceathly R*[2,3], *Thangaraj K*[4,6], *Topf A*[7], *Straub V*[7], *Srivastava P*[1], *Hanna M*[2,3], *Venugopalan Y V*[1]
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59P Clinical and genetic spectrum of GNE myopathy in a neuromuscular cohort in northern India
Rani N[1], *Macken W*[2,3], *Reyaz A*[1], *Ahmad T*[1], *Tarane K*[1], *Danish M*[1], *Consortium ICGNMD-Consortium*[5], *Bhatia R*[1], *Pitceathly R*[2,3], *Thangaraj K*[4,6], *Topf A*[7], *Straub V*[7], *Srivastava P*[1], *Hanna M*[2,3], *Venugopalan Y V*[1]
[1]*All India Institute Of Medical Sciences,* [2]*Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, UK,* [3]*NHS Highly Specialised Service for Rare Mitochondrial Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery,* [4]*Centre for DNA Fingerprinting and Diagnostics,* [5]*https://www.ucl.ac.uk/genomic-medicine-neuromuscular-diseases/global-contributor-list, UCL, UK,* [6]*Centre for Cellular & Molecular Biology,* [7]*John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust*

60P Evaluation of aggrephagy markers in genetically defined MyofibrillarMyopathies

Iannibelli E[1], *Gibertini S*[1], *Maruotti A*[2], *Salerno F*[1], *Cheli M*[3], *Vattemi G*[3], *Tonin P*[3], *Tasca G*[4], *Bortolani S*[5], *Pegoraro E*[6], *Previtali S*[7], *Garibaldi M*[8], *Merlonghi G*[8], *Filosto M*[9,10], *Carnazzi A*[11,1], *Nicolini De Gaetano L*[1], *Riolo G*[1], *Ruggieri A*[1], *Maggi L*[1]

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61P Characteristics of muscle computed tomography in a family with *HSPB8*-related rimmed vacuolar myopathy

Kobayashi M[1], *Abe E*[1], *Wada C*[1], *Yokoyama E*[2], *Hara K*[3], *Nakayama T*[4], *Inoue-Shibui A*[5], *Suzuki N*[6], *Izumi R*[5], *Aoki M*[5], *Nishino I*[7], *Ishihara T*[1], *Toyoshima I*[1]

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62P Axial involvement as a prominent feature in *SMPX*-related distal myopathy

Omar Mohamed Salman D[1], *C. Bolano-Diaz C*[1], *Muni-Lofra R*[1], *Wong K*[1], *Elseed M*[1], *Harris E*[1,2], *Diaz-Manera J*[1], *Guglieri M*[1], *Marini-Bettolo C*[1], *Straub V*[1], *Tasca G*[1]

[1]The John Walton muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, [2]Northern Genetics Service, Institute of Genetic Medicine

63P Identifying the disease-causing variant in a large family, with a late-onset dominant distal myopathy

Turku T[1], *Savarese M*[1], *Johari M*[1,2], *Soininen M*[1], *Hoischen A*[3,4], *Steehouwer M*[3], *Roos A*[5,6], *Preuße C*[7,8], *Stenzel W*[7], *Wallgren-Pettersson C*[1], *Pelin K*[1], *Udd B*[1], *Hackman P*[1]

[1]The Folkhälsan Research Center, [2]Harry Perkins Institute of Medical Research, Centre for Medical Research, [3]Department of Human Genetics, Radboud University Medical Center, [4]Department of Internal Medicine, Radboud Institute for Molecular Life Sciences, Radboud Expertise Center for Immunodeficiency and Autoinflammation and Radboud Center for Infectious Disease (RCI), Radboud University Medical Center, [5]Pediatric Neurology, Faculty of Medicine, University Children's Hospital, University of Duisburg-Essen, [6]Brain and Mind Research Institute, Children's Hospital of Eastern Ontario Research Institute, [7]Department of Neuropathology, Charité-Universitätsmedizin Berlin, Corporate Member of Freie Universität Berlin, Humboldt-Universität Zu Berlin, Berlin Institute of Health (BIH), [8]Department of Neuropediatrics, Charité-Universitätsmedizin Berlin, Corporate Member of Freie Universität Berlin, Humboldt-Universität Zu Berlin, Berlin Institute of Health (BIH)

64P Effects of autophagy stimulation in the Tibial muscular dystrophy mouse model

Sarparanta J[1,2], *Suel-Petat L*[3], *Luque H*[1,2], *Jonson P*[1,2], *Deste G*[4], *Vihola A*[1], *Barresi R*[4], *Udd B*[1,2,5], *Richard I*[3]

[1]Folkhälsan Research Center, [2]Medicum, University of Helsinki, [3]Généthon, [4]San Camillo IRCCS, [5]Neuromuscular Research Center, Tampere University Hospital

65P Striking periscapular muscle hypertrophy as the presenting symptom in a 5-month old infant diagnosed with a *MYH7* congenital myopathy: report of a case

Deconinck N[1], *Coppens S*[2], *Vilain C*[2], *Remiche G*[3], *Semal B*[2], *Presiozi M*[4], *Lecomte S*[5], *Dontaine P*[1]

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66P Clinical and molecular complexity in Desminopathy: description of a novel variant in *DES* gene

Torri F[1], *Torella A*[2], *Ricci G*[1], *Ciurli B*[1], *Piluso G*[2], *Demidov G*[3], *Onore M*[2], *Spampanato C*[2], *Zanobio M*[2], *Rahman S*[2], *Cetrangolo V*[2], *Nigro V*[2], *Siciliano G*[1]

[1]Department of Clinical and Experimental Medicine, University Of Pisa, [2]Department of Precision Medicine, University of Campania "Luigi Vanvitelli", [3]Institute of Medical Genetics and Applied Genomics, University of Tübingen

67P *ADSS1* Myopathy in Korea: a comprehensive analysis of clinical, laboratory, radiological, pathological, and genetic features

Kim S[1], *Choi Y*[1], *Kim W*[2], *Park H*[1], *Choi Y*[1]

[1]Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, [2]Department of Neurology, Kangdong Sacred Heart Hospital, Hallym University College of Medicine,

68P Induced muscle and liver absence of *Gne* in postnatal mice does not result in structural or functional muscle impairment

Mitrani-Rosenbaum S[1], *Harazi A*[1], *Yakovlev L*[1], *Ilouz N*[1], *Selke P*[2], *Horstkorte R*[2], *Argov Z*[3]

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69P Medaka fish Oryzias latipes a new vertebrate model for Titinopathies: the HMERF case study

Cetrangolo V[1,2], *Savarese M*[1,5], *Linari M*[4], *Polishchuk E*[2], *Conte I*[7], *Nigro V*[2,3], *Udd B*[1,5,6]

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70P Defining the landscape of *TIA1* and *SQSTM1* Digenic Myopathy

Fernández-eulate G[1], *Panos-Basterra P*[2], *Theuriet J*[3], *Nadaj-Pakleza A*[4], *Magot A*[5], *Lannes B*[6], *Marcorelles P*[7], *Behin A*[1], *Masingue M*[1], *Caillon F*[8], *Malek Y*[3], *Fenouil T*[9], *Bas J*[10], *Menassa R*[11], *Michel-Calemard L*[11], *Streichenberger N*[9], *Simon J*[12], *Bouhour F*[3], *Evangelista T*[1], *Métay C*[13], *Pegat A*[3], *Stojkovic T*[1]

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71P Phase II/III and efficacy confirmation study of aceneuramic acid for GNE myopathy in Japan

Suzuki N[1,10], *Mori-Yoshimura M*[2], *Katsuno M*[3,4], *Takahashi M*[5], *Yamashita S*[6], *Oya Y*[2], *Hashizume A*[3,4], *Yamada S*[3], *Nakamori M*[5], *Izumi R*[1], *Kato M*[1], *Warita H*[1], *Tateyama M*[1], *Kuroda H*[1], *Asada R*[7], *Yamaguchi T*[8], *Nishino I*[9], *Aoki M*[1]

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72P Skeletal muscle pathology in plectinopathies

Winter L[1]

[1]*Medical University of Vienna*

73P Peripheral Neuropathy in Myofibrillar Myopathies (MFM) and *MFM* gene-related myopathies

Wannarong T[1], *Milone M*[1], *Selcen D*[1], *Dyck P*[1], *Liewluck T*[1]

[1]*Mayo Clinic*

74P Update on GNE-myopathy: introduction of tissue and blood biomarkers and a novel homozygous missense variant associated with early disease onset and proximal involvement

Roos A[1,7,10], *Dobelmann V*[1], *Hentschel A*[2], *Hagenacker T*[3], *Derksen A*[4], *Osmanovic A*[5], *Evangelista T*[6], *Gangfuss A*[7], *Kaiser F*[8], *Schara-Schmidt U*[7], *Ruck T*[1], *Savarese M*[9], *Lochmüller H*[4]

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75P Evaluation of clinico-pathological and genetic spectrum of *FLNC* mutations in Indian cohort

Sharma M[1], *Dhall A*[1], *Jassal B*[1], *Suri V*[1], *Faruq M*[1], *Shamim U*[1], *Bhatia R*[1], *Venugopalan Y V*[1], *Chakarwarti B*[1], *Gulati S*[1]

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76VP Myofibrillar Myopathy with cardiomyopathy, lipid accumulation and sarcoplasmic reticulum dilation

Cotta A[1], *Carvalho E*[1], *da-Cunha-Júnior A*[1], *Menezes M*[1], *da Silveira E*[2], *Costa-e-Silva C*[2], *da-Silva-Neto R*[1], *Cauhi A*[1], *Valicek J*[1], *Vargas A*[1]

[1]*The Sarah Network of Rehabilitation Hospitals,* [2]*The Sarah Network of Rehabilitation Hospitals*

77VP GNE myopathy: Phenotypic diversity in 10-year study of National registry in Japan and correlation of the diaphragm with respiratory function

Yoshioka W[1], *Mori-Yoshimura M*[1], *Oba M*[1], *Saito Y*[1], *Oya Y*[1], *Eura N*[1], *Hayashi S*[1], *Kimura Y*[1], *Sato N*[1], *Nakamura H*[1], *Noguchi S*[1], *Nishino I*[1]

[1]*National Center of Neurology and Psychiatry*

78VP Pathogenic *NARS1* mutations identified as the cause of neurodevelopmental delay, microcephaly and peripheral neuropathy in two related patients

Vlachou V[1], *Abed Alrahman R*[1], *Majumdar A*[1]

[1]*Bristol Royal Hospital for Children*

182P-203P: SMA outcome measures and registries

182P Impact of nusinersen on the health-related quality of life and caregiver burden in patients with Spinal Muscular Atrophy with symptom onset before age 6 months

Lee Y[1], *Bae H*[1], *Shim Y*[2], *Cho J*[3], *Yun J*[4], *Lee H*[5], *Chae J*[6], *Kwon S*[1]

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183P CuidAME: The Spanish longitudinal Registry of SMA patients. A global view in 2024

Nascimento Osorio A[1], *García Uzquiano R*[1], *Puig C*[1], *Fernandez M*[2], *Ñungo N*[3], *Exposito J*[1], *Calvo R*[4], *Alvarez M*[5], *Lopez M*[6], *Martinez E*[7], *Grimalt M*[8], *Fernandez J*[9], *Navarro V*[10], *Urbano M*[11], *Martinez M*[12], *Gonzalez D*[13], *Sariego A*[14], *Sardina M*[15], *Marti I*[16], *CuídAME Study Group*[1]

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184P The integration of PROMs and clinician reported data: a holistic approach to characterise disease burden and treatment impact

Walker H[1], *Page J*[1], *Murphy L*[1], *Segovia S*[1], *Karkkainen E*[1], *Cavalcante E*[2], *Madden M*[2], *Adcock K*[3], *Farrugia M*[4], *Irwin J*[5], *Lilleker J*[6], *McConville J*[7], *Merrison A*[8], *Parton M*[9], *Ryburn L*[10], *Muni-Lofra R*[1], *Scoto M*[2], *Marini-Bettolo C*[1]

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185P Spinal Muscular Atrophy diagnosis in Latin American: The LATAM RegistrAME clinical registry

Carneiro Batista E[1], *Zanoteli E*[2], *Rodrigues Sant'Anna V*[1], *Miranda Duarte F*[1], *de Piano L*[1], *de Pedri E*[1], *de Albuquerque C*[1], *Pedro I*[1], *Serapião A*[1], *Moia D*[1], *Carioca A*[1], *Sampaio B*[1], *Monfardini F*[1], *dos Santos G*[1], *Soares R*[1], *Silva G*[1], *Berwanger O*[3], *Rizzo L*[1], *Fonseca H*[1], *Medical team at the centers T*[4]

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186P Demographic and clinical characteristics of risdiplam-treated and untreated adult patients with SMA

Gorni K[1], *Batech M*[2], *Guittari C*[3], *Carmona A*[2], *Lee S*[4], *Poll A*[4], *Sutherland C*[5], *Marini-Bettolo C*[6], *Walter M*[7], *Jagut M*[8], *Haberlová J*[9], *Hodgkinson V*[10], *Yiu E*[11], *Murphy L*[6], *TREAT-NMD Global Data systems Oversight Committee (TGDOC)*[4], *TREAT-NMD Global Registry Network for SMA*[4], *Gordish-Dressman H*[12], *Simpson A*[5]
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187P French HCPs approach to evaluating SMA adult patients with severe disabilities: a qualitative study

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188P Easy neurocognitive and social evaluation could detect abnormal neurodevelopment in SMA Type 1

Alvarez Molinero M[1], *Pires N*[2], *Garcia Uzquiano R*[2], *Gómez Garcia de la Banda M*[2], *Costa Comellas L*[1], *Ventura Expósito L*[1], *Munell Casadesús F*[1], *Benezit A*[2], *Gómez Andrés D*[1], *Quijano Roy S*[2]
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189P Diagnostic journey of SMA patients in a reference center in Brazil

Rodrigues M[1], *Piauilino Santos Falcão A*[1], *Franco Graça F*[1], *Cavalcanti França M*[1]
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190P Spectrum of phenotypes in SMA patients with four *SMN2* copies in France (Registre SMA France)

Gomez Garcia De La Banda M[1], *Gerin L*[1], *Ropars J*[2], *Garcia-Uzquiano R*[1], *Saugier-Veber P*[3], *Desguerre I*[4], *Salort-Campana E*[5], *Espil C*[6], *Barnerias C*[4], *Laugel V*[7], *Cances C*[8], *Audic F*[5], *Cintas P*[8], *Tard C*[9], *Le Goff L*[10], *Dieterich K*[11], *Drunat S*[12], *Grimaldi L*[13], *Quijano-Roy S*[1]
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191P Characterization of patients SMA diagnosed in Algeria

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192P A community-based experience with the new 12-Tier functional ability scale for Evolving Spinal Muscular Atrophy (EVOLVE-SMA)

Moore Burk M[1,2], *Johnson K*[2,3], *Crawford T*[4], *Apkon S*[1], *Duong T*[5], *Krosschell K*[6], *EVOLVE-SMA Working Group*[1,4,6]
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193P Use of European rare disease registries to describe the natural history and disease progression of Spinal Muscular Atrophy (SMA) over time

Bennett N[1], *Deltour N*[4], *Garry E*[2], *Ferreras A*[4], *Jacquot E*[4], *McElwee S*[2], *Sajedian R*[2], *Kurteva S*[4], *Boin E*[4], *Poll A*[1], *Davies R*[11], *Walter M*[5], *Jagut M*[6], *Haberlová J*[7], *Cattinari M*[8], *Marini-Bettolo C*[9], *Ekström A*[10], *de Lemus Belmonte M*[3], *Breen K*[3], *Servais L*[12]

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194P Longitudinal assessment of the Adapted Test for Neuromuscular Disease (ATEND) in individuals living with Spinal Muscular Atrophy (SMA)

Duong T[1], *Muni-Lofra R*[2], *Pasternak A*[3], *Tang W*[1], *Gu B*[1], *Dunaway Young S*[1], *Wilson A*[7], *Harrington A*[4], *de Monts C*[1], *Jeworek A*[5], *Moore Burke M*[8], *Maczek E*[3], *McIntyre M*[7], *Salvatore S*[1], *Smith S*[1], *Smith R*[4], *Hsu L*[1], *He Z*[1], *Glanzman A*[6], *Day J*[1]

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195P Real-world data on the effectiveness and safety of Onasemnogene abeparvovec in Spinal Muscular Atrophy

Weiß C[1], *Becker L*[1], *Garbade S*[2], *Johannsen J*[3], *Ziegler A*[2]

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196P Profiling neuroinflammatory markers in CSF from paediatric SMA patients in response to nusinersen treatment

Zhang Q[1], *Hong Y*[2], *Brusa C*[3], *Scoto M*[3], *Cornell N*[3], *Patel P*[1], *Boukhloufi I*[3], *Baranello G*[3], *Muntoni F*[3,4], *Zhou H*[1,4]

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197P Survival of SMA type 1 and type 0 infants at the time of disease modifying therapies: results of a 8-year nationwide registry

Coratti G[1], *Pera M*[1], *Agosto C*[2], *D'Amico A*[3], *Ricci F*[4], *Sansone V*[5], *Masson R*[6], *Bruno C*[7], *Varone A*[8], *Pane M*[1], *Mercuri E*[1]

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198P Longitudinal evaluation of fatigue in adult patients with Spinal Muscular Atrophy and the impact of treatment with disease-modifying drugs

Graca F[1], *Iwabe C*[1], *C França Jr. M*[1]

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199P The use of the Motor Unit Number Index (MUNIX) as a biomarker for disease progression in Late-Onset 5q-Spinal Muscular Atrophy treated with Nusinersen

Holanda Mendonca R[1], *Pedro Soares Baima J*[1], *Jorge Polido G*[1], *Otto Heise C*[1], *Zanoteli E*[1]

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200P Minimal detectable change of the Revised Hammersmith Scale in patients with Spinal Muscular Atrophy

O'reilly E[1,2], *Stimpson G*[2], *Rohwer A*[1,2], *Baranello G*[1,2,3], *Muntoni F*[1,2,3], *Scoto M*[1,2,3], *On behalf of SMA REACH UK*[2]

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201P Natural history of Spinal Muscular Atrophy patients with 3 and 4 copies of *SMN2* gene – data from the national Spanish registry (CUIDAME)

Aragon Gawinska K[1], *Fernández García M*[2], *Nascimiento Osorio A*[3], *Paradas C*[4], *Sotoca J*[5], *Povedano M*[6], *Moreno A*[7], *Henao M*[8], *Gil C*[9], *Rojas R*[10], *Gómez Caravaca M*[11], *Grimalt M*[12], *Fernández Torron R*[13], *Jericó I*[14], *García Campos O*[15], *Toledo Bravo de Laguna L*[16], *Hervás D*[17], *Tizzano E*[18], *Vázquez Costa J*[1,19,20], *on behalf of CUIDAME Investigators Group*

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202P Descriptive analysis of the Spinal Muscular Atrophy population treated with Nusinersen included in the CuidAME project

Sotoca Fernandez J[1], *Puig C*[2], *García-Uzquiano R*[2], *García-Romero M*[3], *Pitarch-Castellanos I*[4], *Vázquez-Costa J*[4], *Paradas C*[5], *Povedano M*[6], *Gómez-Caravaca M*[7], *Álvarez-Molinuevo M*[8], *Gómez-Andrés D*[8], *Munell F*[8], *Grimalt M*[9], *Calvo R*[10], *Henao M*[11], *Jericó I*[12], *Fernández-García M*[3]

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203P Towards a better analysis of *SMN2* structures and variants by complete sequencing of the *SMN* locus in an international cohort of 564 SMA patients

Tizzano E[1,2], *Costa-Roger M*[1,2], *Blasco-Pérez L*[1,2], *Tenés A*[1], *Martínez-Cruz D*[1], *Leno-Colorado J*[1,2], *Civit D*[1,2], *Codina-Sola M*[1,2]

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247P-265P, 266VP: Neuromuscular disorders around the world

247P A population-based survey on the knowledge, perception and attitudes towards rare diseases and gene therapy in Belgium: focus on Duchenne Muscular Dystrophy

Van Stappen T[1], *Verschelden M*[1], *Samyn W*[2], *Minnebo J*[2], *Mommen M*[1]

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248P Spinal Muscular Atrophy in Latin American: patient journey observed in regional registry

Carneiro Batista E[1], *Zanoteli E*[2], *Rodrigues Sant'Anna V*[1], *Miranda Duarte F*[1], *de Piano L*[1], *de Pedri E*[1], *de Albuquerque C*[1], *Pedro I*[1], *Serapião A*[1], *Moia D*[1], *Carioca A*[1], *Sampaio B*[1], *Monfardini F*[1], *dos Santos G*[1], *Soares R*[1], *Silva G*[1], *Berwanger O*[3], *Rizzo L*[1], *Fonseca H*[1], *Medical team at the centers T*[4]

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249P Clinical characteristics of the Spinal Muscular Atrophy patients identified in the Brazilian public health system

Batista EC [1], ***Zanoteli E*** [2], Ortega AB[3], Junior MCF[4], .Saute JAM[5], Oliveira ASB.[6], Giannetti JG[7], Pessoa ALS[8], Prufer A[9], Boy R[10], . Junior FGB[1], de Albuquerque CSN[1], Moia DDF[1], Monfardini F[1], Santos GP[1], Soares RVP[1], SilvaGS[1], Rizzo LV[1],. Berwanger O[11], Fonseca HAR[1]

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250P Prevalence of Duchenne Muscular Dystrophy in Italy: a nationwide survey

Cicala G[1,2], Capasso A[1], Ricci M[1], Pane M[1], D'Amico A[3], Bruno C[4], Sansone V[5], Messina S[6], Bello L[7], Masson R[8], Berardinelli A[9], Pini A[10], Ricci F[11], Mongini T[12], Nigro V[13], Comi G[14,15], Battini R[16,17], Arpaia C[1], Coratti G[1], Mercuri on behalf of the Italian DMD group E[1]

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251P Latin-SEQ: a collaborative network to provide genetic diagnosis to patients with neuromuscular diseases in Latin America – project update

Topf A[1], Gonzalez-Chamorro A[1], Laurie S[2,3], Papakonstantinou Ntalis A[2,3], Latin-SEQ Consortium[1], Diaz-Manera J[1]

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252P Natural history and quality of life study on muscular dystrophies in adulthood (Muskel-LIV): study design and conduct

Nordström S[1,2], Wahlgren L[1,2], Lindberg C[1,2], Tulinius M[1], Sofou K[1,2]

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253P International Centre for Genomic Medicine in Neuromuscular Diseases: analysis and characterization of the muscular dystrophy cohort from India

Luce L[1], Vengalil S[3], Nashi S[3], Srivastava K[3], Manoj R[3], Reyaz A[4], Chaudhary N[4], Ahmad T[4], Naveena M[5], Vandrovcova J[2], ICGNMD Consortium, Yareeda S[5], Bhatia R[4], Venugopalan VY[4], Srivastava P[4], Nalini A[3], Töpf A[1], Straub V[1]

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254P "Where can I find a specialized physiotherapist for someone like me?" - A Norwegian innovation project

Ramberg C[1], Hevnskjel Ringvold G[1], Ladehaug T[2], Stokke S[3], Ørstavik K[4], Dybesland Rosenberger A[1]

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255P Relevance of muscle biopsies in the neonatal period: a 52-year retrospective study in the gene-sequencing era

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256P Establishing healthy muscle references and cutoff values for quantitative muscle ultrasound as a foundation for future diagnostic and research use in the neuromuscular clinic

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257P Interim analysis of an integrated interdisciplinary diagnostic pathway in a cohort of unsolved pediatric neuromuscular disorders

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258P Safety and tolerability of Onasemnogene Abeparvovec for patients with Spinal Muscular Atrophy weighing ≤17 kg and ≤24 months old: phase 4 OFELIA study

***Zanoteli E**[1], Muntadas J[2], Gurgel-Gianetti J[3], Monges S[4], Aliberti P[5], Alecu I[6], Ritter S[7], Martins de Lana J[8], Mumneh N[7], Saute J[9,10]*

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259P The 'muscle toolbox': harmonising diagnosis and follow-up for neuromuscular disorders across three Dutch academic hospitals

***Cameron D**[1], van Doorn J[1], Heskamp L[2], Rauh S[3], Becks R[1], Kruit M[3], Nievelstein R[2], Tromp S[3], Tannemaat M[3], Braakman H[1], Erasmus C[1], Houwen-van Opstal S[1], Niks E[3], van der Pol L[2], Kan H[3], Froeling M[2], Bartels B[2], van Alfen N[1]*

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260P NEUROMYODredger – 3 billion megaproject: expanding the accessibility of exome sequencing for the diagnosis of neurodevelopmental and neuromuscular disorders in nine countries

***Malfatti E**[1], Caramizaru A[2], Lee H[3], Kim J[3], Shoaito H[4], Pennisi A[1], Fahmi N[5], Escobar-Cedillo R[6], Miranda-Duarte A[6], Nouioua S[7], Benchaabi O[7], Martinez P[8], Castiglioni C[9], Dobrescu A[2], Tajsharghi H[10]*

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261P Insights into Facioscapulohumeral dystrophy in African individuals: clinical and molecular findings from a collaborative study

***Rodriguez Cruz P**[1,2,3], Diagne R[2,3], Henning F[4], Naidu K[4], Heckmann J[5], Floudiotis N[5], Malfatti E[6], Kamissoko Y[7], Leturcq F[8], Urtizberea A[9], Tellez M[10], Elsheikh B[10], Beltran S[1], Diop A[2,3], Ndiaye M[2,3], Hodes R[11], Voermans N[12], Van der Vliet P[13], Van der Maarel S[13], Lemmers R[13]*

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262P A global analysis of the CMT1A locus: implications for the origin and susceptibility to Charcot- Marie-Tooth disease type 1A across populations

***Rodriguez Cruz P**[1,2,3], Alitsiou A[1], Diagne R[2,3], Lia-Baldini A[4], Ghorab K[5], Gallo Diop A[2,3], Ndiaye M[2,3], Beltran S[1], Lao O[7]*

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263P Updated genetic testing in heart-transplant recipients in Norway between 1983-2022 uncovering genes relevant to neuromuscular disorders

Benterud A[1,2], *Haug Popperud T*[1,3], *Broch K*[4], *Hasselberg N*[4], *Prøven Bogsrud M*[5], *Berge K*[5], *Haugaa K*[4,6], *Ørstavik K*[1,3]

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264VP A nationwide register study on the epidemiology of Duchenne Muscular Dystrophy (DMD) in Finland

Auranen M[1], *Kyttälä M*[2], *Vesikansa A*[3], *Mehtälä J*[3], *Isohanni P*[4,5], *Kosunen M*[2]

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265P Effective supplementation of 6'-sialyllactose in GNE Myopathy: the results from a placebo-controlled, pilot study

Park Y[1,2], *Kim D*[2,3], *Choi J*[4], *Jung J*[4], *Kim L*[4], *Shin J*[2,3]

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266VP Cardiovascular comorbidities in Myasthenia Gravis: a systematic review and meta-analysis

Tahir S[1], *Iguh C*[1], *Sadik O*[2], *Duggirala N*[3], *Ezenwa V*[1], *Ahmed S*[1]

[1]*Windsor University School of Medicine,* [2]*Jackson Park Hospital,* [3]*Nri Medical College*

280P-315P, 316VP-317VP: Dystrophinopathies (animals models, biomarkers, brain, genetics)

280P Technology-enabled assessment of cognition to facilitate democratization of brain health in Duchenne Muscular Dystrophy

Thangarajh M[1]

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281P Prognostic significance of *ACTN3* genotype in Duchenne Muscular Dystrophy: findings from an Argentine patient cohort

Luce L[1,2,3], *Mazzanti C*[1,2], *Carcione M*[1,2], *Llames Massini C*[1,2], *Buonfiglio P*[4], *Dalamón V*[4], *Bolaño Díaz C*[3,5], *Mesa L*[5], *Dubrovsky A*[5], *Cotignola J*[6,7], *Giliberto F*[1,2]

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282P Biomarker panel for Duchenne Muscular Dystrophy clinical trials

Tsioutsias I[1], *Bautista A*[1], *Terrill J*[1], *Bakker A*[1], *Pinniger G*[1], *Arthur P*[1]

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283P Generation of cardiomyocyte cell models for personalized medicine

Nhi P[1], *Åstrand C*[1], *Hedhammar M*[1], *Al-Khalili Szigyarto C*[1]

[1]*KTH Royal Institute of Technology*

284P Suspension bead array profiling of 1018 serum proteins in search for disease progression biomarkers for Duchenne Muscular Dystrophy

Jiménez-Requena Carrasco A[1], *Ajeenah A*[1], *Johansson C*[1], *Naveed A*[5], *Tobin R*[2], *Degan C*[3], *de Vries S*[3], *van der Burgt Y*[3], *Diaz-Manera J*[4], *Guglieri M*[4], *CINRG-DNHS Investigators C, for DMD investigators, Dang U*[2], *Tsonaka R*[3], *Spitali P*[3], *Hathout Y*[5], *Al-Khalili Szigyarto C*[1]

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285P AAV-delivered U7snRNA restores full-length dystrophin in patient cell lines with DMD intronic pseudoexon mutations

Beljan J[1], *Gushchina L*[1], *Lin H*[1], *Nicolau S*[1,2,3], *Weiss R*[4], *Flanigan K*[1,2,3]

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286P Cognitive deficits in Duchenne muscular dystrophy: Analysis of assessment, mutation location, and social vulnerability

Arun S[1], *Kaat A*[2], *Thangarajh M*[1]

[1]*Virginia Commonwealth University*, [2]*Northwestern University*

287P Localisation of Dystrophin isoforms in the mouse brain: insights into neuropsychiatric comorbidities in Duchenne Muscular Dystrophy

Aghaeipour A[1,2,3], *Tetorou K*[1,2], *Gileadi T*[1,2], *Morgan J*[1,2], *Montanaro F*[1,2], *Muntoni F*[1,2,3]

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288P In patients with Duchenne Muscular Dystrophy fragmented QRS in the lateral lead of the electrocardiography is associated with reduced cardiac function

Yamamoto T[1], *Ogawa S*[1], *Nambu Y*[2], *Bo R*[2], *Matsuo M*[3], *Awano H*[4]

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289P Non-tandem duplications in DMD: impacts on genetic counseling and medical decision making

Gross B[1], *Higginbotham E*[2], *Lau L*[2], *Sung W*[2], *Moran O*[3], *Hasnain A*[2,4], *Stavropoulos D*[2,4], *Beaulieu Bergeron M*[5,6], *Boycott K*[7,8], *McNiven V*[9], *Liu R*[10], *Matesanz S*[1,11]

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290P Developing a non-invasive, in vivo method for evaluating muscle inflammation longitudinally in two mouse models of Duchenne Muscular Dystrophy

MacKinnon A[1], *Rowsell J*[1], *Uaesoontrachoon K*[1], *Mullen A*[1], *Hoffman E*[1,2], *Nagaraju K*[1,2]

[1]*Agada Biosciences Inc.*, [2]*School of Pharmacy and Pharmaceutical Sciences, Binghamton University*

291P Novel insights into the expression and the epigenetic modulation of LKB1, a potential diagnostic and therapeutic player in Duchenne muscular *dystrophy*

Boccanegra B[1], *Mantuano P*[1], *Conte E*[1], *Tulimiero L*[1], *Lenti R*[1], *Quarta R*[1], *Cristiano E*[1], *Decio A*[2], *Cappellari O*[1], *Licandro S*[2], *Fossati G*[2], *Steinkühler C*[2], *De Luca A*[1]

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292P Phenotypic characterization of the D2-mdx mouse for Duchenne Muscular Dystrophy: updates from a natural history study

Tulimiero L[1], *Mantuano P*[1], *Boccanegra B*[1], *Lenti R*[1], *Quarta R*[1], *Cristiano E*[1], *Marinelli M*[1], *Mele A*[1], *Cappellari O*[1], *De Luca A*[1]

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293P Evaluation of creatine/creatinine ratio and myostatin as monitoring biomarkers for DMD patients using real-world data

Degan C[1], *Vries S*[1], *Ikelaar N*[1], *Tsonaka R*[1], *Niks E*[1,2], *Spitali P*[1,2]

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294P Identification of disease-specific extracellular vesicle-associated plasma biomarkers for Duchenne muscular dystrophy (DMD) and Facioscapulohumeral Muscular Dystrophy (FSHD)

Bayazit M[1], *Henderson D*[3], *Tawil R*[3], *Flanigan K*[1,2], *Harper S*[1,2], ***Saad N***[1,2]

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295P Diagnostic yield of next-generation sequencing compared with muscle biopsy for diagnosis of MLPA negative Duchenne Muscular Dystrophy

Sanmaneechai O[1], *Meedee M*[1], *Pimchanok K*[1]

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296P Development of monitoring biomarker signatures in Duchenne Muscular Dystrophy

De Vries S[1], *Degan C*[1], *Tobin R*[2], *Jimenéz-Requena A*[3], *Ajeenah A*[3], *Johansson C*[3], *Van der Burgt Y*[1], *Diaz-Manera J*[4], *Guglieri M*[4], *Al-Khalili Szigyarto C*[3], *Spitali P*[1], *Hathout Y*[5], *Tsonaka R*[1], *Dang U*[2], *on behalf of Team: CINRG-DNHS Investigators, on behalf of Team: DMD Investigators*

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297P In-depth behavioral characterization of Duchenne Muscular Dystrophy mouse models lacking one, multiple or all dystrophin isoforms in the brain

Van Putten M[1], *Verhaeg M*[1], *van der Pijl L*[1], *van de Vijver D*[1], *Tanganyika-de Winter C*[1], *Stan T*[1], *Aartsma-Rus A*[1]

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298P Magnetic resonance imaging analyses of the brain of Duchenne Muscular Dystrophy mouse models lacking one, multiple or all dystrophin isoforms

Van Putten M[1], *Verhaeg M*[1], *Suidgeest E*[1], *Kan H*[1], *van der Weerd L*[1], *Aartsma-Rus A*[1]

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299P Detailed natural history study of the D2-mdx and BL10-mdx models for Duchenne Muscular Dystrophy

Van Putten M[1], *Mantuano P*[2], *Boccanegra B*[2], *Tanganyika-de Winter C*[1], *Putker K*[1], *Schneider A*[1], *Tulimiero L*[2], *Mele A*[2], *van de Vijver D*[1], *Gordiss-Dressman H*[3], *Cappellari O*[2], *Aartsma-Rus A*[1], *De Luca A*[2]

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300P Brain involvement in Duchenne Muscular Dystrophy (BIND)

Tetorou K[1,2], *Aghaeipour A*[1,2], *Gileadi T*[1,2], *Ma S*[1,2], *Perdomo Quinteiro P*[3], *Saoudi A*[5,6], *Ceschi L*[5], *Zarrouki F*[5], *Mitsogiannis M*[7], *Fergus C*[9], *Kelly V*[9], *Zhang L*[4], *Spitali P*[3], *Aoki Y*[8], *Morgan J*[1,2], *Montanaro F*[1,2], *Vaillend C*[5], *Goyenvalle A*[6], *Muntoni F*[1,2]

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301P Interactome analysis of dystrophin isoforms in the mouse brain

Tetorou K[1], *Aghaeipour A*[1,2], *Gileadi T*[1,2], *Pablo P*[3], *Zhang L*[4], *Spitali P*[3], *Morgan J*[1,2], *Montanaro F*[1,2], *Muntoni F*[1,2]

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302P Dystrophin quantification by capillary western immunoassay and western blot across the dystrophinopathy spectrum

Lay J[1], *Flanigan K*[1,2,3], *Nicolau S*[1,2,3]

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303P A longitudinal study on cognitive and behavioural functioning in Duchenne Muscular Dystrophy

Govaarts R[1], *Weerkamp P*[2], *de Vreede M*[1], *Marini-Bettolo C*[3], *Geagan C*[3], *Hollingsworth K*[4], *Hendriksen J*[2,5], *Kan H*[1,5], *Niks E*[1,5], *Straub V*[3]

[1]*Leiden University Medical Center,* [2]*Kempenhaeghe,* [3]*Newcastle University and Newcastle Hospitals NHS Foundation Trust,* [4]*Newcastle University,* [5]*Duchenne Centre*

304P Multi-modal magnetic resonance imaging protocols in the multi-site Brain Involvement in Dystrophinopathies (BIND) study

Govaarts R[1], *Doorenweerd N*[1,2], *Brogna C*[4], *Clark C*[3], *Guliaeva I*[3], *Hollingsworth K*[5], *MacDonald Fisher P*[6], *Mercuri E*[4], *Niks E*[1], *Parikh J*[5], *Seunarine K*[3], *Smythe L*[3], *Stemmerik M*[6], *Straub V*[2], *Verdolotti T*[4], *Vissing J*[6], *Würgler Slipsager A*[6], *Muntoni F*[3], *Kan H*[1], *Kerkelä L*[3]

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305P Investigation of the clinical significance of serum titin in Duchenne Muscular Dystrophy

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306P Evaluating genetic therapies targeting the central nervous system in Duchenne Muscular Dystrophy using high-resolution spatial transcriptomics

***Mao Q**[1], Ahmadi A[1], van Doorne L[1], de Vries S[1], Heezen L[1], Vacca O[2], Aartsma-Rus A[1], van Putten M[1], Goyenvalle A[2], Mahfouz A[1,3,4], Spitali P[1]*

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307P Uncovering the genomic basis of cognitive comorbidities in Duchenne Muscular Dystrophy using spatial transcriptomics in mouse models

***Mao Q**[1], Heezen L[1], de Vries S[1], van Doorne L[1], Aartsma-Rus A[1], van Putten M[1], Mahfouz A[1,2,3], Spitali P[1]*

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308P Nephrological parameters in boys with Muscular Dystrophy

***Rohlenová M**[1], Zieg J[2], Lauerová B[1], Kumhera M[1], Gloser M[1], Dolanská A[1], Haberlová J[1]*

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309P Cholesterol defects in Muscular Dystrophy patients and mice cause ambulation dysfunction: insight into statin- and anti-HMGCoAR-induced myopathies

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310P Stress exacerbates glucose intolerance in the mdx mouse model of Duchenne Muscular Dystrophy

***Lindsay A**[1,2,3], Salimova E[4,5], Eliades J[4], Zheng G[4], Caeyenberghs K[3], de Veer M[4,5]*

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311P Development and pilot validation of the DuMAND checklist to screen for Duchenne Muscular Dystrophy-Associated Neurobehavioral Difficulties (DuMAND)

***Geuens S**[1,2], Goemans N[1], Lemiere J[3,4], Doorenweerd N[5], De Waele L[1,2]*

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312P Genotype and corticosteroid treatment are associated with distinctive variations in gray matter characteristics among patients with Duchenne Muscular Dystrophy

***Geuens S**[1,2], Van Dessel J[3], Kan H[4,5], Govaarts R[4,5], Niks E[5,6], Goemans N[1], Lemiere J[7,8], Doorenweerd N[4], De Waele L[1,2]*

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313P Validation lab: allowing standardized in vitro and in vivo experiments for candidate treatments for Duchenne Muscular Dystrophy

***Stan T**[1], Van De Vijver D[1], Tanganyika-de Winter C[1], van der Pijl L[1], Aartsma-Rus A[1]*

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314P Protein biomarkers predicting clinical milestones in Duchenne Muscular Dystrophy combining natural history and real-world data

*Spitali P[1,2], Ikelaar N[2,3], Barnard A[4], Eng S[5], Hosseini Vajargah S[5], Ha K[5], Kan H[2,6], Vandenborne K[4], Niks E[2,3], **Walter G**[7]*

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315P Cardio-metabolic and cytoskeletal proteomic signatures differentiate fear sensitivity in dystrophin-deficient mdx mice

Major G[1], *Herbold C*[1], *Cheng F*[2], *Lee A*[2], *Russell A*[3], *Lindsay A*[1,3,4]

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316VP What you see is what it is: The tales of two brothers with rare intronic dystrophin gene duplication

Khries M[1], *Gowda V*[1]

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317VP Overview of a cohort of 35 manifesting dystrophinopathy females – and addressing the forthcoming challenges

Goncalves A[1,2,3], *Sousa A*[3,4], *Garrido C*[3,5], *Cardoso M*[3,4], *Pinto M*[3,6], *Vieira E*[1,2], *Oliveira M*[1,2], *Taipa R*[2,3,6], *Coelho T*[3,4], *Santos M*[3,5], *Santos R*[1,2,3]

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438P - 440P, 442P - 471P: Myotonic dystrophy

438P Knockdown of *DMPK* in Myotonic Dystrophy Type 1 using CRISPR Cas13

Oh H[1,2], *Todorow V*[1], *Hintze S*[1], *Schoser B*[1], *Meinke P*[1,2]

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439P Cardiac abnormalities and management in children with myotonic dystrophy type 1

Bovenkerk D[1], *Van Den Akker R*[2], *Klinkenberg S*[1], *Louw J*[3], *Udink Ten Cate F*[4], *Braakman H*[2]

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440P Nonclinical data for PGN-EDODM1 demonstrated nuclear delivery, mechanistic and meaningful activity for the potential treatment of DM1

Gilbert J[1], *Klein A*[2], *Lonkar P*[1], *Gutnick A*[1], *Foy J*[1], *Yu S*[1], *Reid T*[3], *Sarkar K*[3], *Cleary J*[3], *Berglund J*[3], *Furling D*[2], ***Holland A***[1]

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442P Cognitive diversity in Congenital Myotonic Dystrophy: implications for early intervention

Dixon M[1], *Butterfield R*[1]

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443P Assessing inter and intra-rater reliability of video hand opening time in Myotonic Dystrophy

Mcintyre M[1], *de Monts C*[2], *Wilson A*[1], *Massey C*[3,4], *Olson S*[1], *Nelson L*[5], *Hageman N*[2], *Tang W*[2], *Dekdeburn J*[6], *Duong T*[2]

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444P Exploring Hand Myotonia: assessing hand opening and individual finger movements through machine learning

Duong T[1], *de Monts C*[1], *McIntyre M*[3], *Karatsidis A*[2], *Juraver A*[2], *Ataide P*[1], *Hageman N*[1], *Erb K*[2], *Meilleur K*[2], *Day J*[1], *Burton L*[2], *Kanzler C*[2]

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445P Utilization and costs of healthcare services and labour market affiliation of persons with adult-onset Myotonic Dystrophy – a Danish Register-Based Study (Study II)

Handberg C[1,2], *Rudolfsen J*[3], *Andersen H*[4], *Vissing J*[5], *Rossau C*[6], *Dreyer P*[6], *Olsen J*[3], *Bengtsson S*[3], *Aagaard H*[1], *Werlauff U*[1]

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446P Comorbidities and mortality of persons with adult-onset Myotonic Dystrophy – a Danish Register-Based Study (Study I)

Werlauff U[7], *Rudolfsen J*[2], *Andersen H*[3], *Vissing J*[4], *Rossau C*[5], *Dreyer P*[6], *Olsen J*[2], *Bengtsson S*[2], *Aagaard H*[7], *Handberg C*[6,7]

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447P Atrio-ventricular conduction in a mouse model with Myotonic Dystrophy Type-1: a preliminary study in hDMPK-77Tg mice

Kimura K[1], *Minegishi K*[2], *Motohashi N*[2], *Morita H*[3], *Nakanishi K*[3], *Daimon M*[3], *Takeda N*[3], *Echigoya Y*[4], *Nakamori M*[5], *Aoki Y*[2]

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448P Quantitative analysis of brain atrophy in patients with myotonic dystrophy (DM1) using MRI -multi center analysis

Nakayama T[1], *Matsumura T*[2], *Kuru S*[3], *Kobayashi M*[4], *Suwazono S*[5], *Takahashi M*[6]

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449P Molecular biomarkers in myotonic dystrophy type 1

Slipsager A[1], *Hildonen M*[2], *Godtfeldt Stemmerik M*[1], *Tümer Z*[2], *Dunø M*[2], *Birkedal U*[2], *Vissing J*[1]

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450P A review of the symptom control clinic, advance care planning and mortality in myotonic dystrophy 1 in a UK neuromuscular centre

Willis T[1,2], *Bassie C*[1], *Fox H*[3], *Kulshrestha R*[1], *Willis D*[2,3]

[1]*Robert Jones and Agnes Hunt hospital,* [2]*University of Chester Medical School,* [3]*Severn Hospice*

451P Gait parameters / cognitive function correlation in a cohort of patients with myotonic dystrophy type 1: a single-center sensor-based gait analysis

Risi B[1], *Pilotto A*[2,3,4], *Rizzardi A*[2,3], *Ferrari E*[1], *Labella B*[2,3], *Zatti C*[2,3], *Hansen C*[5], *Romijnders R*[5], *Caria F*[1], *Damioli S*[1], *Bertella E*[1], *Poli L*[3], *Ferullo L*[2,3], *Olivieri E*[2,3], *Maetzler W*[5], *Padovani A*[2,3], *Filosto M*[1,2]

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452P Genetic confirmation of myotonic dystrophy type II after allogeneic stem cell transplant

Schaefer J[1], *Saak A*[2]

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453P Characterisation of cell culture models of myotonic dystrophy type I for drug screening by in-cell western and digital droplet PCR

López-Martínez A[1], *Sendino M*[1], *Torres-Conde N*[1], *Al-Ani A*[1], *Martínez-Gonzalez S*[1], *Nuñez-Manchón J*[2], *Nogales-Gadea G*[2], ***Arechavala Gomeza V***[1,3]

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454P Myotonic dystrophy type 1 with progressive supranuclear palsy showing responsiveness to levodopa

Indrawati L[1,2], *Tunjungsari D*[1,2,3], *Nagpal C*[2], *Muljono W*[1], *Ariarini N*[2,3]

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455P Psoriasis and myotonic dystrophy type 1: another cutaneous manifestation of a multisystemic disorder

Tufano L[1], *Bucci E*[1], *Antonini G*[1], *Garibaldi M*[1]

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456P Gait analysis by IMU sensor in myotonic dystrophy type 1

Tufano L[1], *Bianchini E*[1], *Bucci E*[1], *Antonini G*[1], *Garibaldi M*[1]
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457P Mortality rate and predictors of death in the DM1 population, a registry-based study

Bassez G[1], *Kachal A*[1], *Gyenge M*[1], *Hamroun D*[1], *French Myotonic Dystrophy Study Group*
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458P The iDM-Scope Registry: an innovative France-Canada framework to advance Myotonic Dystrophy translational research

Bassez G[1], *Gyenge M*[1], *Hamroun D*[2], *Kachal A*[1], *Evangelista T*[1], *Rodrigue X*[3], *Nury M*[3], *Lochmüller H*[4], *Gagnon C*[5]
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459P Correlation of minimally invasive blood biomarkers with muscle-derived molecular signatures in myotonic dystrophy type 2

Kleefeld F[1], *Hentschel A*[5], *Preusse C*[1], *Schoser B*[3], *Stenzel W*[1], *Roos A*[2,5,7]
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460P Understanding the clinical heterogeneity in myotonic dystrophy type 1: identifying clinical phenotypes using unsupervised clustering

la Fontaine L[1], *Imkamp M*[1], *'t Hoen P*[2], *van As D*[2], *Smulders F*[2], *Bruijnes J*[1], *de Kok J*[1], *Faber C*[1], *Merkies I*[1], *van Kuijk S*[1]
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461P Evaluation of PGN-EDODM1: FREEDOM-DM1 and FREEDOM2-DM1 clinical trials in myotonic dystrophy type 1

Larkindale J[1], *Shoskes J*[1], *Garg B*[1], *Song G*[1], *Lonkar P*[1], *Babcock S*[1], *Vacca S*[1], *Yu S*[1], *Mellion M*[1]
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462P Evaluating EEG as an outcome measure for CNS Symptoms in myotonic dystrophy type 1: a clinical trial analysis

Allen S[1], *Kamali T*[1], *Parker D*[1], *Seto A*[2], *Ehrich E*[2], *Wang E*[3], *Sampson J*[1]
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463P Longitudinal progression of motor function in individuals with myotonic dystrophy type 1: insights from the END-DM1 study

Johnson N[1], *Hung M*[1], *Sansone V*[1], *Subramony S*[1], *Statland J*[1], *Hamel J*[1], *van Engelen B*[1], *Mul K*[1], *Elsheikh B*[1], *Roxburgh R*[1], *Day J*[1], *Swenson A*[1], *Schoser B*[1], *Ragole T*[1], *Greene E*[1], *Shieh P*[1], *Thornton C*[1], *On behalf of DMCRN*
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464P Advancing preclinical research of myotonic dystrophy type 1 with 3D functional human skeletal muscle tissues

Fernández-Garibay X[1], *Sabater-Arcís M*[2,3], *Núñez-Manchón J*[4], *Tejedera-Villafranca A*[1], *Artero R*[2,3], *Suelves M*[4], *Nogales-Gadea G*[4], *Ramón-Azcón J*[1,5], *Fernández-Costa J*[1]
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465P Factors impacting dysphagia in adult-onset myotonic dystrophy type 1

Berggren K[1], *Kamarunas E*[2], *Johnson N*[1]
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466P Multisystemic manifestations in adult patients with classic myotonic dystrophy Type 1

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467P A multicenter retrospective study in Turkish children with myotonic dystrophy type 1

Oz Tuncer G[1], *Sanri A*[2], *Kurt Bayir G*[1], *Erol İ*[3], *Ardıçlı D*[4], *Öztürk M*[5], *Gazeteci Tekin H*[6], *Kutluk G*[7], *Hergüner Ö*[8], *Tekgül H*[9], *Tütüncü Toker R*[10], *Per H*[11], *Çavuşoğlu D*[12], *Pembegül Yildiz E*[13], *Kömür M*[14], *Türkdoğan D*[15], *Özgör B*[16], *Ayça S*[17], *Turkish Myotonic Dystrophy Type 1 Study Group T*[18]

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468P Exploring gut microbiome alteration in children with myotonic dystrophy type 1: a pilot study

van Uden A[1,2], *Zweers-van Essen H*[3], *de Baaij-Daalmeyer A*[3], *Ederveen T*[2], *Braakman H*[1]

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469P Modal allele change as a predictor of skeletal muscle symptoms progression in myotonic dystrophy type 1

Radovanovic N[1], *Pesovic J*[1], *Peric S*[2,3], *Radenkovic L*[1], *Brkusanin M*[1], *Brajuskovic G*[1], *Rakocevic Stojanovic V*[2,3], *Savic-Pavicevic D*[1]

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470P Splicing is improved using a novel AAV-microRNA delivery platform as a treatment for myotonic dystrophy type 1

Cao S[1], *Tomassy G*[1], *Richards B*[1], *Fan W*[1], *Luo Z*[1], *Jackson R*[1], *O'Riordan C*[1], *Goulet M*[1], *Mueller C*[1]

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471P Interfering with CUG toxic repeats using AAV.U7snRNAs rescue myotonia and splicing defects in myotonic dystrophy type 1

Almeida C[1], *Brinkman A*[1], *Wendt C*[2], *Blatnik A*[2], *Delgado A*[1], *Gushchina L*[1], *Arnold W*[3], *Weiss R*[4], *Wein N*[1,5]

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598P-618P: Registries, networks and care of NMD

598P The Canadian Neuromuscular Disease Registry: using real-world evidence to further spinal muscular atrophy research in Canada

Crone M[1], *Henley K*[2], *Hodgkinson V*[2], *Dyck A*[2], *Brais B*[3], *Campbell C*[4], *Gonorazky H*[5], *Lochmüller H*[6], *MacKenzie A*[7], *McMillan H*[7], *Oskoui M*[8], *Korngut L*[9], *Selby K*[10], *SMA Investigator Network on behalf of the CNDR Investigator Network*

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599P The UK Facioscapulohumeral muscular dystrophy Patient Registry: a powerful tool to support clinical research and patient voice in the translational research pathway

Walker H[1], *Muni-Lofra R*[1], *Orrell R*[2], *Graham A*[3], *Norwood F*[4], *Roberts M*[5], *Willis T*[6], *Matthews E*[7], *Mencias M*[7], *Adcock K*[8], *Marini-Bettolo C*[1]

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600P The UK Myotonic Dystrophy Patient Registry - empowering clinical research and patient voice with an effective translational research tool

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

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601P Gap analysis of 4-year data in the Dutch Dystrophinopathy Database

Meijer-Krom Y[1,3], *Ikelaar N*[1,3], *Bongers J*[1,3], *van de Velden N*[1,3], *Snijder R*[4], *Houwen van Opstal S*[2,3], *Niks E*[1,3]

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602P FISMA for high quality and interoperable real-world data on dystrophinopathies: FAIR from the start, from concept to reality

Meijer-Krom Y[1,5], *Hoek R*[1,5], *Houwen van Opstal S*[2,5], *van de Velden N*[1,5], *Ikelaar N*[1,5], *van der Holst M*[3,5], *Snijder R*[4], *Niks E*[1,5]

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603P DMD Hub: An established clinical trial accelerator network delivering tools and services to sites, patients and industry in the UK

Heslop E[1], *Gaeta A*[2], *Reuben E*[2], *Johnson A*[2], *Cammish P*[1], *McNiff M*[1], *Riguzzi P*[1], *Muntoni F*[3], *Childs A*[4], *Straub V*[1], *Guglieri M*[1]

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604P Descriptive analysis of Duchenne Muscular Dystrophy patients included in the Swedish National Registry for neuromuscular disorders

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605P Dutch LGMD registry: real-world data to facilitate trial rea*diness*

Schrama E[1], *Hoek R*[1], *Bongers J*[1], *Straathof C*[1], *Badrising U*[1], *van Reenen R*[2], *van der Kooi A*[3], *van Duyvenvoorde H*[4], *Krom Y*[1], *Niks E*[1]

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606P Burden of illness for male patients with Duchenne Muscular Dystrophy in a real-world setting, a Swedish registry study

Furby H[1], *Boudreau D*[2], *Ekstrom A*[3], *Freilich J*[4], *Kroksmark A*[5], *Streja E*[4], *Wojtowicz J*[1], *Yang Q*[4], *Zhao J*[4], *De Ford C*[1]

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607P A patient-centered registry for rare neuromuscular disorders with federated FAIR infrastructure: the EURO-NMD Registry Hub

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608P Collection of real-world data for Duchenne Muscular Dystrophy patients through a national registry: description of the current cohort in the UK

Brooke M[1], *Wolfe A[1,2], Aslam N[1], Manzur A[2], Muntoni F[1,2], Baranello G[1,2]*

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609P Integration of healthcare and research to optimize treatment and collect real-world data in neuromuscular disorders

Verschoof M[1,2], *Slingerland F[1], Roest A[3], de Vreede I[3], Warmenhoven N[4,5], van Beek C[5], de Witte P[5], Bot D[6], van Wageningen H[4,5], Atsma D[7], Straathof C[1,2], Kan H[8], Spitali P[2,9], Snijder R[10], Hoek R[1], van der Holst M[2,5], Meijer-Krom Y[1,2], Niks E[1,2]*

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610P Facilitating patient/caregiver preparation prior to visit increases quality in neuromuscular care

Edofsson U[1], *Weichbrodt J[1], Sofou K[1]*

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611P Empowering patient decision-making and enhancing clinical care through the UK Duchenne Muscular Dystrophy patient data collection platform

Reuben E[1], *Gaeta A[1], Johnson A[1], Philippault H[1], Davies E[2], Ferrer-Mallol E[2], Cammish P[3], Guglieri M[3], Ezzamouri B[1]*

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612P Dystrophinopathy in ACTION; Analysis of the first 500 males enrolled in the Advanced Cardiac Therapies Improving Outcomes Network prospective dystrophinopathy registry

Hayes E[1], *Villa C*[2], *Nandi D*[1], *Soslow J*[3], *Mokshagundam D*[4], *Shih R*[5], *Wisotzkey B*[6], *Parent J*[7], *Cunningham T*[8], *Conway J*[9], *Esteso P*[10], *Birnbaum B*[11], *Shugh S*[12], *Raucci F*[13], *Kaufman B*[14], *Soares N*[15], *Kirmani S*[16], *Martinez H*[17], *Gambetta K*[18], *Lal A*[19], *Wittlieb-Weber C*[20]

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613P Contemporary use of ventricular assist devices in Muscular Dystrophy: a report from the ACTION learning collaborative

Nandi D[1], *Hayes E*[1], *Auerbach S*[2], *Bansal N*[3], *Conway J*[4], *Esteo P*[5], *Kaufman B*[6], *Lal A*[7], *Law S*[8], *Mokshagundam D*[9], *Sutcliffe D*[10], *Absi M*[11], *Raskin A*[12], *Friedland-Little J*[13], *Wittlieb-Weber C*[14], *Kroslowitz B*[15], *Lorts A*[15], *Cripe L*[1], *Villa C*[15]

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614P Burden on parent caregivers of children with a muscle disease: a qualitative study

Altena-rensen S[1], *van Hattum M*[2], *Braakman H*[1], *Erasmus C*[1],

[1]*Radboudumc Amalia Children's Hospital, Netherlands,* [2]*Hogeschool Arnhem en Nijmegen*

615P Introducing PaLaDIn: improving the use of rare NMD patient data to inform healthcare decision making

Leary R[1], *Allison D*[2], *Cogoni S*[3], *Cornet R*[4], *Roos M*[5], *Davies E*[6], *Schoser B*[7], *Kahtava K*[8], *Ezzamouri B*[9], *Straub V*[1]

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616P From New Zealand Neuromuscular Disease Registry to Pūnaha Io - the New Zealand NeuroGenetic Registry & Biobank

Rodrigues M[1,2], *O'Grady G*[3], *Buchanan C*[1], *Fraser L*[1], *Stewart C*[1], *Cleland J*[4], *Roxburgh R*[1,2]

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617P Using innovative data modelling methods to improve data quality- learning from Adult SMA REACH a real-world data collection study

Segovia Simon S[1], *Verdu-Diaz J*[1], *Karkkainen E*[1], *Page J*[1], *Carver A*[1], *Alvarez G*[1], *Muni-Lofra R*[1], *Mitchell-Sodhi J*[1], *Marini-Bettolo C*[1]

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618P Global FKRP Registry - the research database for Limb Girdle Muscular Dystrophy 2I/R9

McDonald S[1], *Murphy L*[1], *Alfano L*[2], *Brazzo K*[3], *Johnson N*[4], *Laurent J*[5], *Mathews K*[6], *Thiele S*[7], *Vissing J*[8], *Walter M*[7], *Woods L*[9], *Ørstavik K*[10], *Straub V*[1]

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15:15-15:45	**Short Oral Presentations 7**	**Short Oral Presentations 8**	**Short Oral Presentations 9**
	North Hall	Terrace 2A	Terrace 2B
	69P, 70P, 74P, 71P, 72P	**469P, 470P, 471P, 468P, 203P**	**315P, 314P, 312P, 265P, 262P**
	Moderator: Duygu Selcen, The Mayo Clinic	*Moderator: Nicholas Johnson, Virginia Commonwealth University*	*Moderator: Anna Sarkozy, Great Ormond Street Hospital*

15:45-16:45 **Poster Session 4** Forum Hall (refreshments provided)

98P-109P, 110VP-112VP: ALS/neuropathy

98P Biallelic pathogenic PLEKHG5 variants in a girl with childhood-onset lower motor neuron disease

Çavuşoğlu D[1], *Ataseven Kulali M*[2], *Guzel A*[3], *Olgac Dundar N*[4]
[1]*Departments of Pediatric Neurology, Afyonkarahisar Health Sciences University,* [2]*Departments of Pediatric Genetics, Etlik City Hospital,* [3]*Departments of Neurology, Afyonkarahisar Health Sciences University,* [4]*Department of Pediatric Neurology, Faculty of Medicine, İzmir Katip Celebi University*

99P Genetic odyssey: a journey through inherited neuropathies in our center

Davarasingi V[1], *Krishna G*[1], *Ramesh babu R*[1], *Satyam P*[1], *Mathew A*[1]
[1]*Synapse Neuro center and Child Development Centre*

100P Case series of three children with *SH3TC2*-Related Hereditary Sensorimotor Neuropathy

Moore Burk M[1], *Kelley C*[1], *Silver C*[1], *Gibbons M*[2], *Foster H*[1], *Browning K*[1,2], *Murphy-Zane M*[1,2], *Apkon S*[1,2], *Yang M*[1,2]
[1]*Children's Hospital Colorado,* [2]*University of Colorado School of Medicine*

101P Pediatric Charcot-Marie-Tooth (CMT) Clinic: quality care and preparing for the future

Moore Burk M1, *Kelley C*[1], *Silver C*[1], *Gibbons M*[2], *Foster H*[1], *Browning K*[2], *Murphy-Zane M*[1,2], *Yang M*[1,2], *Apkon S*[1,2]
[1]*Children's Hospital Colorado,* [2]*University of Colorado School of Medicine*

102P Clinical profiles bulbar and limb onset Amyotrophic Lateral Sclerosis patients: A Retrospective Cohort Study

Yunisova G[1], *Üçem S*[2], *Özdağ Acarlı A*[3], *Başak A*[4], *Oflazer P*[3]
[1]*Koc University Hospital,* [2]*Koç University, Koç University Hospital, Department of Neurology,* [3]*Koç University, Koç University Hospital, Department of Neurology, Muscle Diseases Center,* [4]*Koç University, Research Center for Translational Medicine (KUTTAM), NDAL, Department of Molecular Biology and Genetics*

103P NMD670, a first-in-class skeletal muscle ClC-1 Inhibitor in Charcot-Marie-Tooth disease: the SYNAPSE-CMT phase 2 study

Gidaro T[1], *S. Grønnebæk T*[1], *Cornwall C*[1], *Gupte J*[1], *Sampson C*[1], *Kiyasova V*[1], *H. Pedersen T*[1], *A. Quiroz J*[1]
[1]*NMD Pharma*

104P RNA Aptamers as a potential drug candidate for ALS

Niu L[1], *Huang Z*[1], *Akamatsu M*[1]
[1]*Department of Chemistry, University at Albany, State University of New York*

105P Frequency and clinical characterization of *SORD*-Related Neuropathy in a Belgian cohort

Opsomer M[1,2], *Vermeersch P*[3], *Philip V*[1], *Decru B*[3], *Race V*[4], *Dohrn M*[5,6], *Zuchner S*[5], *Claeys K*[1,2]
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106P Clinical, neurophysiological, and genetic characterization of a 5' UTR PSAP variant: Implications for diagnosis

Donkervoort S[1], *Orbach R*[1], *Silverstein S*[1,2], *Potticary A*[1], *Stenton S*[3], *Karachunski P*[4], *Bönnemann C*[1]
[1]*NNDCS, National Institutes of Health,* [2]*Rutgers New Jersey Medical School,* [3]*Broad Institute of MIT and Harvard,* [4]*University of Minnesota Medical School*

107P Clinical features of FOSMN syndrome in Korea: a comparison with bulbar-onset Amyotrophic Lateral Sclerosis

Choi S[1], *Ju W*[1], *Sung J*[1]
[1]*Department of Neurology, Seoul National University Hospital*

108P Case series: description of *HSBP1* mutations conferring motor neuropathies with overlap features presenting barriers to timely diagnosis

Nardin J[1], *Segall H*[1], *Jones K*[2], *Mehrabyan A*[1]
[1]*UNC Health, 2Duke University*

109P Preserve sense by antisense – a novel allele-specific antisense oligonucleotide therapy for *SPTLC1*-related Hereditary Sensory Neuropathy

*Meng J[1,2], Ma S[1], Wong L[1], Lone M[3], Zhang Q[1], Cheng S[1], Demetriou C[1], Clark A[4], Bennett D[5], Hornemann T[3], Reilly M[1], Muntoni F[1,2], **Zhou H**[1,2]*

[1]University College London, [2]NIHR Great Ormond Street Hospital Biomedical Research Centre, [3]University of Zürich, [4]Queen Mary University, [5]University of Oxford

110VP *FIG4* loss of function: Unraveling the phenotypic spectrum for clinical trial readiness

Boyek G[1], *Orbach R[1], Gottlieb K[2], Duff J[3], Lenk G[4], Bönnemann C[1]*

[1]National Institutes of Health, Neuromuscular and Neurogenetic Disorders of Childhood Section, [2]Elpida Therapeutics, [3]CureCMT4J/Talia Duff Foundation, [4]University of Michigan, Department of Human Genetics

111P Genotype-phenotype analysis of multisystem proteinopathy with neurological involvement

Xia X[1], *Dong Y[1], Chen Y[1], Zhao C[1], Zhu W[1]*

[1]Huashan Hospital, Fudan University

112VP Spinal and Bulbar Muscular Atrophy with easy fatigability and hyperCKemia

Lee J[1]

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148P-181P: SMA Therapies

148P Rapid risdiplam initiation in newborns with Spinal Muscular Atrophy (SMA): A multicenter, retrospective cohort study

Goedeker N[1], *Dierker A[1], Felker M[2], Lakhotia A[3], Rogers A[3], Zaidman C[1]*

[1]Washington University in Saint Louis School of Medicine, [2]Indiana University School of Medicine, [3]University of Louisville School of Medicine

149VP 3D stem cell-derived spinal cord/muscle organoid model for studying and treating neuromuscular diseases

Corti S[1], *D' Angelo A[1], Beatrice F[1], Ongaro J[2], Rinchetti P[1], Faravelli I[1], Miotto M[3], Lodato S[3], Nizzardo M[2], Comi G[1], Ottoboni L[1]*

[1]University Of Milan, [2]Fondazione IRCCS Ca Granda Ospedale Maggiore Policlinico, [3]Humanitas University

150P Patient, caregiver and healthcare professionals' perspective on spinal bracing in early onset Spinal Muscular Atrophy in the U.K.: a national survey

Quelch W[1], *Taylor F[2], Thorman P[3], Ramjattan H[2], Ramdas S[2,4,5], Ong M[1]*

[1]Department of Paediatric Neurology, Sheffield Children's Hospital, [2]Oxford University Hospitals NHS Foundation Trust, [3]Spinal Muscular Atrophy UK, [4]Department of Paediatric Neurology, John Radcliffe Hospital, [5]MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford

151P Real-world treatment with risdiplam in adults with SMA: a multicentre study

Gorni K[1], *Guittari C[2], Candrilli S[3], Miles L[3], Simpson A[4], Shapouri S[5]*

[1]PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, [2]PDMA Neuroscience and Rare Disease, Genentech, Inc., [3]RTI Health Solutions, [4]Global Access, F. Hoffmann-La Roche Ltd, 5Genentech, Inc.

152P Perceived effects of treatments by SMA adult patients: a French qualitative study

Laforêt P[24,2,3], *Montagu G[4], Boyer F[5,6], Gargiulo M[7,8], Pouplin S[9,10], Barrière A[11], Berling E[1,2], Bonnyaud C[9,10], Cintas P[12], Hogrel J[7], Le Goff L[13], Marchadier B[14], N'Dah Sékou G[1,2], Orlikowski D[2,15,16,17], Prigent H[2,17,18], Ropars J[19,23], Salort-Campana E[20,21], Stojkovic T[7,22], Attarian S[20,21]*

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153P Long-term effectiveness of risdiplam in non-ambulant SMA patients with prolonged disease duration

Weng W[1,2], *Shieh J*[1], *Huang H*[1], *Tsai L*[1]
[1]*National Taiwan University Hospital,* [2]*National Taiwan University College of Medicine*

154VP Usage of disease modifying therapies in Spinal Muscular Atrophy and existing disparities: a population-based study from the MDA MOVR database

McLaren N[1], *Joo D*[1], *Nowak R*[1], ***Roy B***[1]
[1]*Yale University School of Medicine*

155P The experience of parents as they make treatment decisions for their child with Spinal Muscular Atrophy and the factors that influence these decisions: a narrative review

Leach M[1], *Finanger E*[1], *Izumi S*[1]
[1]*Oregon Health and Science University*

156P Phosphorylated neurofilament heavy chain in cerebrospinal fluid and plasma in clinically silent and childhood-onset SMA individuals from Serbia

Brkusanin M[1], *Kosac A*[2], *Brankovic-Sreckovic V*[2], *Jovanovic K*[3], *Karanovic J*[1], *Matijasevic Jokovic S*[1], *Garai N*[1], *Pesovic J*[1], *Radovanovic N*[1], *Radenkovic L*[1], *Dobrijevic Z*[4], *Nikolic D*[3,5], *Stevic Z*[5,6], *Brajuskovic G*[1], *Milic-Rasic V*[2,5], *Savic-Pavicevic D*[1]
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157P Functional progression post Nusinersen therapy in Spinal Muscular Atrophy Type 2: a comprehensive 18-month analysis from a South Indian centre

Banavara Shyamprasad S[1], *Khandekar G*[1], *Ramesh Babu R*[1,2], *Maganthi M*[2], *Krishna G*[1], *Kumar A*[2], *Mathew A*[1,2]
[1]*Synapse Neuro Center, Bengaluru, India,* [2]*Bangalore Baptist Hospital*

158P Results of the nationwide pilot project of newborn screening for SMA in Czech Republic

Gloser M[1], *Lauerova B*[1], *Rohlenova M*[1], *Kumhera M*[1], *Dolanska A*[1], *Jurikova L*[2], *Haberlova J*[1]
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159P Acute liver failure failure post gene therapy in Spinal Muscular Atrophy

Satyam P[1,2], *Krishna G*[1,2], *Ramesh Babu R*[1,2], *Maganthi M*[2], *K.S L*[2], *H.R S*[3], *Mathew A*[1,2]
[1]*Synapse Neuro and Child Development Center,* [2]*Bangalore Baptist Hospital,* [3]*Gleneagles Global Hospital*

160P Risdiplam real-world experience in paediatric patients with Spinal Muscular Atrophy type 2

Nascimento Osorio A[1], *Expósito-Escudero J*[1], *Medina J*[1], *Valle M*[1], *Roca S*[1], *Moya O*[1], *Natera De-Benito D*[1], *Latre C*[1], *Ortez C*[1], *Carrera-García L*[1]
[1]*Hospital Sant Joan De Deu*

161P Long-term safety of Onasemnogene Abeparvovec for patients with Spinal Muscular Atrophy: real-world findings from the RESTORE Registry

Servais L[1,2], *Alecu I*[3], *Lopez-Leon S*[4], *Ilic A*[3], *Brueckner A*[3], *Benguerba K*[5], *Reyna S*[6], *Dabbous O*[6], *Mumneh N*[6], *Finkel R*[7]
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162P Real-world outcomes following Onasemnogene Abeparvovec in patients with SMA and Invasive Ventilatory support: findings from the RESTORE Registry

Servais L[1,2], *Shieh P*[3], *Goedeker N*[4], *Waldrop M*[5,6], *Bo R*[7], *Erbas Y*[8], *Raju D*[9], *Benguerba K*[10], *Reyna S*[9], *Wolff D*[9], *Finkel R*[11]
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163P Real-World outcomes following Onasemnogene Abeparvovec in patients with SMA and One SMN2 gene copy: findings from the RESTORE Registry

Servais L[1,2], *Mathews K*[3], *Bernes S*[4], *Lakhotia A*[5], *Tizzano E*[6], *Raju D*[7], *Benguerba K*[8], *Reyna S*[7], *Dabbous O*[7], *Finkel R*[9]

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164P Meta-analysis of clinical impact of Onasemnogene Abeparvovec treatment of Spinal Muscular Atrophy patients

Reyna S[1], *Dabbous O*[1], *Wallach S*[1], *Patel A*[1], *Toro W*[1], *Ritter S*[1]

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165P Health Care Resource Use (HCRU) for patients with Spinal Muscular Atrophy (SMA) Types 2 or 3 in the UK

Reyna S[1], *Toro W*[1], *Patel A*[1], *Saleh S*[2], *Dabbous O*[1]

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166P Fertility outcomes in risdiplam-treated male patients with Spinal Muscular Atrophy (SMA): a multicenter case series

Erdler M[1], *Coskery S*[2], *Frey M*[3], *Lopez M*[4]

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167P SnRNAseq analysis of muscle samples of patients with SMA reveals novel pathogenic pathways and open avenues for new therapeutic strategies

Collins C[1], *Gokul-Nath R*[1], *Katsikis P*[1], *Fernández-Simón E*[1], *Villalobos E*[1], *Monceau A*[1], *Reza M*[1], *Mehra P*[1], *Laidler Z*[1], *Clark J*[1], *Rojas-García R*[2], *Tasca G*[1], *Marini-Bettolo C*[1], ***Diaz-Manera J***[1]

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168P Long-term effects of risdiplam in adult Spinal Muscular Atrophy: a 24-month prospective study on clinical, functional, and patient-reported outcome measures

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169P Alteration of LARGE1 abundance in patients and a mouse model of 5q-associated Spinal Muscular Atrophy

Roos A[1,4,5], *Schmitt L*[2], *Hansmann C*[2], *Hezel S*[2], *Salmanian S*[2], *Hentschel A*[3], *Meyer N*[1], *Della Marina A*[1], *Kölbel H*[1], *Kleinschnitz C*[2], *Schara-Schmidt U*[1], *Leo M*[2], *Hagenacker T*[2]

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170P Apitegromab in Spinal Muscular Atrophy: baseline characteristics of participants enrolled in the phase 3 SAPPHIRE study

Crawford T[1], *Servais L*[2], *Krueger J*[3], *Kölbel H*[4], *Gomez Garcia M*[5,6,7], *Cances C*[8], *Kuntz N*[9,10], *Finkel R*[11], *Yao B*[12], *Zhao G*[12], *Marantz J*[12], *Darras B*[13], *Mercuri E*[14,15]

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171P Newborn Spinal Muscular Atrophy screening: 2-year results of multi-center experience in Turkey

Ünver O[1], *Öztürk G*[1], *Sakarya Güneş A*[2], *Özçelik A*[3], *Maraş Genç H*[4], *Saltık S*[5], *Akbeyaz İ*[1], *Tekin Orgun L*[2], *Karakayalı B*[1], *Acar Arslan E*[2], *Cırdı G*[2], *Ceylan A*[6], *Gökdemir Y*[7], *Özbaş Akkaya A*[8], *Erdem Eralp E*[7], *Karadağ Saygı E*[9], *Karadağ B*[7], *Kara B*[2], *Topaloğlu H*[10], *Türkdoğan D*[1]

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172P FUS protein expression in the myopathology of 5q-associated Spinal Muscular Atrophy type 3

Kölbel H[1], *Dobelman V*[2], *van Haute L*[3], *Lancene E*[4], *Kollipara L*[5], *Della Marina A*[1], *Horvath R*[6], *Schara-Schmidt U*[1], *Ruck T*[2], *Schoser B*[7], *Evangelista T*[4], *Roos A*[1,2,8]

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173P A multidisciplinary approach to address the challenges posed by new phenotypes of symptomatic SMA patients treated with innovative therapies

Gaume M[1], *Viallard L*[2], *Vialle R*[1], *Desguerre I*[3], *de Lattre C*[4], *Cunin V*[5], ***Vuillerot C***[6]

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174P Risdiplam in type 2 and 3 Spinal Muscular Atrophy: results of a cohort of adult Italian patients. A 3-year follow-up

Ruggiero L[1], *Russo A*[1], *Iodice R*[1], *Dubbioso R*[1], *Tozza S*[1], *Esposito G*[2], *Zoppi D*[1]

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175P Thrombospondin-4 as potential cerebrospinal fluid biomarker for therapy response in pediatric 5q-associated spinal muscular atrophy patients

Dobelmann V[1], *Roos A*[1,2,3], *Hentschel A*[4], *Della Marina A*[2], *Leo M*[5], *Schmitt L*[5], *Maggi L*[6], *Schara-Schmidt U*[2], *Hagenacker T*[5], *Ruck T*[1], *Koelbel H*[2]

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176P Acceptability, feasibility, safety and potential efficacy of an optimised rehabilitation for treated patients with SMA in UK: ACE SMA

Lilien C[1], *Hill S*[1], *Mavrommati F*[1,2], *Ramjattan H*[3], *Taylor F*[3], *Collett J*[2], *Servais L*[1,4]

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177P Effect of gene therapy on the epigenetic and transcriptional stability of SMA

Smeriglio P[1], *Grandi F*[1], *Arnould A*[1], *Pezet S*[1], *Marais T*[1], *Mazzucchi S*[1], *Astord S*[1], *Cohen-Tannoudji M*[1]

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178P Spinal muscular atrophy type II skeletal muscle treated with Nusinersen and Risdiplam shows SMN restoration but mitochondrial deficiency

Grandi F[1], *Astord S*[1], *Pezet S*[1], *Gidaja E*[1], *Mazzucchi S*[1], *Chapart M*[1], *Vasseur S*[1], *Mamchaoui K*[1], *Smeriglio P*[1]

[1]*INSERM UMR 974*

179P Intravenous and intrathecal Onasemnogene Abeparvovec gene therapy in symptomatic and presymptomatic SMA: long-term follow-up study

Darras B[1], *Farrar M*[2,3], *Mercuri E*[4], *Strauss K*[5,6,7], *Day J*[8], *Chien Y*[9], *Masson R*[10], *Bernardo R*[11], *Alecu I*[12], *Dodd N*[13], *Mehl L*[14], *Connolly A*[15,16]

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180P Interim results from RESPOND: a study of nusinersen in children with Spinal Muscular Atrophy (SMA) previously treated with onasemnogene abeparvovec (OA)

Masson R[1], *Proud C*[2], *Finkel R*[3], *Parsons J*[4], *Kuntz N*[5], *Foster R*[6], *Li W*[7], *Chary S*[7], *Sohn J*[7], *Fradette S*[7], *Youn B*[7], *Paradis A*[7]

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181P Investigating the developmental expression of the SMN protein in brain in a Spinal muscular atrophy mouse model

Demetriou C[1], *Frontzek K*[3], *Zhou H*[2], *Muntoni F*[1], *Baranello G*[1]

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234P-246P, 497P-513P, 514VP-515VP: Myasthenia Gravis, NMJ1-2, Periodic paralysis

234P Congenital Myasthenic Syndromes in adults: clinical features, diagnosis and long-term prognosis

Theuriet J[1,2], *Villar-Quiles R*[2], *Stojkovic T*[2], *Eymard B*[2]

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235P Cross sectional study of 187 patients with Congenital Myasthenia Syndrome, describing the clinical phenotypes, genetic mutations, and single point standardised assessment scores

Ramdas S[1,2], *Ramjattan H*[3], *Hennehan L*[4], *Natera De Benito D*[5], *Nascimiento A*[5], *Della Marina A*[6], *Schara-Schmidt U*[6], *Munot P*[7], *Simmons E*[7], *Maggi L*[8], *Gallone A*[9], *Nadaj Pakleza A*[10], *Chanson J*[10], *Marini-Bettolo C*[11], *Moat D*[11], *Paz Guerrero-Molina M*[12], *Dominguez-González C*[12,13], *Jungbluth H*[14,15], *Sheehan J*[14], *Palace J*[4]

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236P Real-world experience with C5 complement inhibition and FcRn modulation in Myasthenia Gravis

Huntemann N[1], *Nelke C*[1], *Schroeter C*[1], *Meuth S*[1], *Ruck T*[1]

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237P Advancing the understanding of VAMP1-related Congenital Myasthenic Syndrome: phenotypic insights, favorable response to 3,4-diaminopyridine, and clinical characterization of five new cases

Natera De Benito D[1], *Pugliese A*[2], *Polavarapu K*[3], *Guergueltcheva V*[4], *Tournev I*[5], *Todorova A*[6], *Ribeiro J*[7], *M Fernández-Mayoralas D*[8], *Ortez C*[1], *Martorell L*[1], *Estevez-Arias B*[1], *Matalonga L*[9], *Laurie S*[9], *Jou C*[1], *Lau J*[3], *Thompson R*[3], *Shen X*[10], *Engel A*[10], *Nascimento A*[1], *Lochmuller H*[3], *Selcen D*[10]

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238P NMD670, a first-in-class skeletal muscle ClC-1 Inhibitor in Myasthenia Gravis: the SYNAPSE-MG dose-finding study

Gidaro T[1], *S. Grønnebæk T*[1], *Cornwall C*[1], *Gupte J*[1], *Sampson C*[1], *Kiyasova V*[1], *H. Pedersen T*[1], *A. Quiroz J*[1]

[1]*NMD Pharma A/S, DK-8200*

239P The association between disease severity and utilities, mental health, fatigue, sleep disturbances and sick leave in patients with Myasthenia Gravis

Brackx F[1], *Goffart B*[2], *Linthoudt T*[2], *De Ruyck F*[2], *Phillips G*[2], *Dewilde S*[1], ***Claeys K***[3,4]

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240P Late presentation *DOK7* Congenital Myasthenia Syndrome (CMS), misdiagnosed as Myasthenia Gravis (MG)– a case series

Henehan L[1], *Rossini E*[1,2], *Dong Y*[3], *Beeson D*[4], *Ramdas S*[5,6], *Leite I*[1,4], *Palace J*[1,4]

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241P Long term follow-up of CHRNE Congenital Myasthenic Syndrome (CMS) – a retrospective multi-centre cohort study

Henehan L[1], *Khries M*[2], *Ramjattan H*[3,4], *Dong Y*[5], *Everett R*[1], *Munot P*[6], *Jungbluth H*[2,7], *Beeson D*[8], *Ramdas S*[3,9], *Palace J*[1,8]

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242P Efgartigimod in AChR and non-AChR generalized Myasthenia Gravis: a single center experience

Antozzi C[1], *Frangiamore R*[1], *Rinaldi E*[1], *Vanoli F*, *Bonanno S*[1], *Maggi L*[1], *Mantegazza R*[1]

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243P The effect of Nav1.4 Ile582Val gain-of-function mutation on mouse skeletal muscle excitability is sex specific

Suetterlin K[1,2,3], *Mannikko R*[2], *Matthews E*[5], *Maitland S*[4], *Greensmith L*[2], *Hanna M*[2], *Tan S*[2,6]

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244P A natural history study of Congenital Myasthenic Syndromes, to establish reliable outcome measures suitable for clinical and research assessment

Ramjattan H[1], *English H*[1], *Hennehan L*[2], *Ramdas S*[3,4], *Palace J*[2]

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245P Exploring the relationship between an instrumented walking test and community physical activity in individuals with Congenital Myasthenic Syndrome

Ramjattan H[1], English H[1], Ramdas S[2,3], Esser P[4], Palace J[5]

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246P Factors affecting the diagnostic delay of Myasthenia Gravis

Marlet I[1], Andersen R[1], Axelsen K[1], Andersen L[1], Wissing J[1], Witting N[1]

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497P AChR-positive generalized Myasthenia Gravis patients unresponsive to new targeted molecular therapies: a single-centre case series

Rossini E[1,2], Marando D[1], Morino S[2], Leonardi L[2], Tufano L[1], Lauletta A[1], Forcina F[1], Garibaldi M[1,2], Antonini G[1], Fionda L[2]

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498P Neuromuscular centre effect on prevalence of Congenital Myasthenic Syndrome (CMS) in the UK

Rossini E[1,2], Henehan L[1], Everett R[1], Dong Y[3], Marini Bettolo C[4], Munot P[5], Jungbluth H[6], Beeson D[7], Ramdas S[8,9], Palace J[1,7]

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499P Exploring the impact of diet and physical activity on episodic muscle weakness and paralysis in HypokalemicPeriodicParalysis: a qualitative interview study

Welland N[1,2], Hagen Venås B[1,2], Ellefsen-Martinsen M[1], Ludt Fossmo H[1,3,4], Dybesland Rosenberger A[5], Dahl H[6], Ørstavik K[3], Nordstrøm M[1,3]

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500P The 6-minute walk test in a small group of patients with Congenital Myasthenic Syndromes

Bulut N[1], Aydın Yağcıoğlu G[2], Demir A[3], Gürbüz İ[1], Yılmaz Ö[1], Haliloğlu G[3]

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501P Validation of ME&MG, a novel digital device for patients with Generalized myasthenia Gravis: the DOMYA Study

Berling E[1], Orlikowski D[1], Nicolas G[2], Prigent H[2], N'Dah-Sekou G[2], Carment L[3], Gorin C[3], Klaeyle L[3], Touré Cuq E[3], Aras E[4], Zinaï S[3], ***Laforêt P***[2]

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502P Cognitive functioning in Myasthenia Gravis: an Italian cohort study

Marcassoli A[1], Raggi A[1], Lanza M[1], Curatoli C[1], Fornari A[1], Leonardi M[1], Maggi L[2], Bonanno S[2], Mantegazza R[2], Antozzi C[2], Vanoli F[2], Frangiamore R[2]

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503P Corticosteroid dose tapering in patients with generalised Myasthenia Gravis on zilucoplan: an interim analysis of RAISE-XT

Boroojerdi B[1], *Freimer M*[2], *Genge A*[3], *Hewamadduma C*[4,5], *Leite M*[6], *Beau Lejdstrom R*[7], *Grimson F*[8], *Savic N*[7], *Howard Jr. J*[9]

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504P Myonuclear abnormalities in Hypokalemic Periodic Paralysis due to ATP1A2 variant

Yae Y[1,2], *Ogasawara M*[1,3], *Nonaka I*[1], *Hayashi S*[1], *Iida A*[4], *Okamura-Oho Y*[5], *Noguchi S*[1], *Nishino I*[1,4]

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505VP Safety and effectiveness of ravulizumab in gMG: evidence from a global registry

Narayanaswami P[1], *Macwan S*[2], *Winkley J*[3], *Gordon A*[4], *Pulley M*[5], *Greene E*[6], *Zeinali L*[7], *Rodrigues E*[7], *Yegin A*[7], *Howard Jr J*[8]

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506VP Long-term efficacy and safety of ravulizumab in adults with AChR-Ab+ gMG: final results from the phase 3 CHAMPION MG open-label extension

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507P Late-onset and milder phenotypes in a *DOK7* Myasthenic Syndrome cohort to rethink the natural history of a perceived severe disease

Brisson J[1,2,3,4], *Villeneuve J*[5], *Nicolas L*[3], *Mathieu J*[1,2], *Gagnon C*[1,3,4]

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508P Clinical and genetic features of patients with Myotonia Congenita in Korea

Choi Y[1], *Kim S*[1], *Hong J*[2], *Park H*[1], *Choi Y*[1]

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509P A real-life experience with Eculizumab and Efgartigimod in generalized Myasthenia Gravis patients

Fionda L[1], *Pane C*[2], *Di Stefano V*[3], *Cuomo N*[2], *Sarnataro A*[2], *Vinciguerra C*[4], *Bevilacqua L*[4], *Brighina F*[3], *Rini N*[3], *Puorro G*[2], *Marsili A*[2], *Garibaldi M*[5], *Saccà F*[2]

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510P Eculizumab versus rituximab for refractory anti-acetylcholine receptor antibody-positive generalized Myasthenia Gravis: a single-center experience

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

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511P Blood biomarkers in a cohort of patients with CHRNE-associated Congenital Myasthenic Syndrome

Della Marina A[1], Koutsoulidou A[2], Natera de Benito D[3], Tykocinski L[4], Tomazou M[5], Georgiou K[2], Kölbel H[1], Nascimento A[3], Ortez C[3], Lochmüller H[6,7,8,9], Phylactou L[2], Ruck T[10], Abicht A[11,12], Schara-Schmidt U[1], Kale D[13], Hentschel A[13], Roos A[1,6,10]

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512P Investigating muscle specific kinase agonist antibody treatment for Agrn and ColQ Congenital Myasthenic Syndromes

Ho K[1,2], Adjei-Afriyie O[1,2], Carmona R[1], Ray R[2], Zeldin J[1,2], O'Neil D[1], Vanhauwaert R[4], Spendiff S[1], Lochmüller H[1,2,3]

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513P Deposition of MAC and IgG subclasses at the neuromuscular junction in LRP4+ Myasthenia Gravis

Hoffmann S[1], Meisel A[1], Brokamp K[1], Helmig L[1], Schülke-Gerstenfeld M[2], Rückert J[3], Pumberger M[4], Schömig F[4], Ruck T[5], Pawlitzki M[5], Stenzel W[6], ***Preusse C***[1,2,6]

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514VP Latent tuberculosis infection in Myasthenia Gravis patients in a low-incidence regio*n*

Carbonero C[1], Arias M[2], Mozo L[3], Palacios J[4], ***Moris De La Tassa G***[1]

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515VP *SCN4A* loss of function zebra fish model: insights into myopathy and sudden infant death

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318P-358P, 343VP, 359VP-360VP: DMD - clinical care and cases reports, BMD

318P DMD splice site point mutation in intron 68 maintains the reading frame and almost full-length dystrophin expression with cell-specific pattern

Białobrzeska M[1], Przymuszała M[1], Potulska-Chromik A[2], Kostera-Pruszczyk A[2], Stępniewski J[1], Florczyk-Soluch U[1], Dulak J[1]

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319P MRI gluteal/thigh muscle fat fraction detects NSAD motor task failures in men with Becker muscular dystrophy

Rock K[1], Willcocks R[1], Barnard A[1], Forbes S[1], Lott D[1], Senesac C[1], Rooney W[2], Baetscher E[2], Subramony S[1], Chahin N[1], Kamal O[1], Hinkle J[1], Corwine A[1], Huerta O[2], Walter G[1], Vandenborne K[1]

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320P Clinical characterization of a large single-centre cohort of patients with Becker Muscular Dystrophy

Riguzzi P[1,2], Borland H[1], Bourke J[1], Marini Bettolo C[1], James M[1], Muni Lofra R[1], Diaz-Manera J[1], Tasca G[1], Elseed M[1], Grover E[1], Geagan C[1], Schiava M[1], Bolano Diaz C[1], Michell-Sodhi J[1], Moat D[1], Wong K[1], Pegoraro E[2], Bello L[2], Straub V[1], Guglieri M[1]

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321P A new world: proposed model for physical therapist evaluation and management following implementation of newborn screening for Duchenne Muscular Dystrophy

Smith M[1], *Iammarino M*[1], *Reash N*[1], *Steiner C*[1], *Chagat S*[2], *Meyer A*[3,1], *Alfano L*[1,4], *Lowes L*[1,4]

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322P Rasch evaluation of North Star Ambulatory Assessment and North Star Assessment for Limb-Girdle type Muscular Dystrophies in Becker Muscular Dystrophy

Lowes L[1], *James M*[2], *Alfano L*[1], *Mayhew A*[2], *Eagle M*[3], *McDonald C*[4], *Vissing J*[5], *Phan H*[6], *Donovan J*[7], *MacDougall J*[7]

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323P Caregiver burden with Duchenne and Becker Muscular Dystrophy in Japan

Ishizaki M[1], *Kobayashi M*[2], *Nakamura A*[3], *Maeda Y*[1], *Ueyama H*[1], *Matsumura T*[4]

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324P A longitudinal study of creatine kinase, creatinine levels, and appendicular lean mass index in pediatric Becker Muscular Dystrophy

Horn P[1,2], *Zygmunt A*[1,2], *Vilaisaktipakorn P*[1], *Sharaf T*[1,3], *Shiuan Y*[2], *Rybalsky I*[1], *Reebals L*[1], *Bange J*[1], *Tian C*[1,2]

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325P Is upper extremity contracture progression related to changes in upper extremity function in Duchenne Muscular Dystrophy? An international multicenter natural history study

Van Der Holst M[1,2], *Italianer M*[2], *Wolfe A*[3], *Chesshyre M*[4], *Voit T*[5], *Straub V*[6], *de Lucia S*[7], *Servais L*[8,9], *Hogrel J*[7], *Pelsma M*[10], *de Groot I*[10], *Houwen S*[10], *Muntoni F*[3], *Niks E*[1]

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326P Acute adverse events after Zoledronic acid infusion are uncommon in patients with Duchenne Muscular Dystrophy previously treated with oral bisphosphonates

Vilaisaktipakorn P[1], *Nasomyont N*[1,2], *Hornung L*[1,2], *Tian C*[1,2], *Wasserman H*[1,2], *Rutter M*[1,2]

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327P Correlation of biomarkers and timed function tests in pediatric Becker Muscular Dystrophy

Vilaisaktipakorn P[1], *Zygmunt A*[1,2], *Sharaf T*[3], *Shiuan Y*[2], *Horn P*[1,2], *Bange J*[1], *Reebals L*[1], *Rybalsky I*[1], *Tian C*[1,2]

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328P Navigating Dystrophinopathies: dual diagnosis and their implications

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329P Siblings with Duchenne Muscular Dystrophy: a chart review to explore associations between age of diagnosis and clinical disease outcomes

Bhimarao Nagaraj C[1,2], *Brahmamdam V*[1,2], *Pilipenko V*[1], *Armstrong N*[3], *Zygmunt A*[1,2], *Rybalsky I*[1,2], *Reebals L*[1], *Tian C*[1,2]

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330P Trends in creatine kinase levels and 10-meter walk-run velocity in pediatric Becker Muscular Dystrophy treated with glucocorticoid steroids

Zygmunt A[1,2], *Vilaisaktipakorn P*[1], *Sharaf T*[1,3], *Shiuan Y*[2], *Horn P*[1,2], *Bange J*[1], *Reebals L*[1], *Rybalsky I*[1], *Tian C*[1,2]

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331P A Phase 1/2 clinical trial of the antisense oligonucleotide BMN 351 now actively recruiting boys with exon 51–skip-amenable Duchenne Muscular Dystrophy

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[1]BioMarin Pharmaceutical Inc

332P Physician and caregiver concordance in Duchenne Muscular Dystrophy patients in Europe, Japan, and the USA: a multi-national survey

***Posner N**[1], Talaga A[1], Strober J[3], Ishigaki K[2], Cappelleri J[1], Dukacz S[1], Aslam Z[1], Morton E[4], Iqbal H[4], Chatterton E[4], DeCourcy J[4]*
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333P Real-World symptom progression in Duchenne Muscular Dystrophy patients in Europe, Japan, and the USA: results from a multi-national survey

***Posner N**[1], Talaga A[1], Strober J[3], Ishigaki K[2], Cappelleri J[1], Dukacz S[1], Aslam Z[1], Morton E[4], Iqbal H[4], Chatterton E[4], DeCourcy J[4]*
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334P GrowDMD: an international study on transition of youth with Duchenne Muscular Dystrophy (DMD)

***Friedrich S**[1], Langer T[1], Reeskau G[2], Rodger S[1], Willems J[1], de Angelis F[3], Brigladori B[4], Guastafierro E[4], Leonardi M[4], Marcassoli A[4], Moroni I[4], Nardocci N[4], Fournier A[5], Frei J[6], Gutierrez Rojas R[5], Kraus De Camargo O[6], Pozniak K[6], Swain A[6], Gorter J[6], Osman H[7]*
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335P Transition in Duchenne Muscular Dystrophy: understanding healthcare providers' roles in an international context

***Friedrich S**[1], Langer T[1], Reeskau G[2], Rodger S[1], Willems J[1], de Angelis F[3], Brigladori B[4], Guastafierro E[4], Leonardi M[4], Marcassoli A[4], Moroni I[4], Nardocci N[4], Fournier A[5], Frei J[6], Gutierrez Rojas R[5], Kraus De Camargo O[6], Pozniak K[6], Swain A[6], Gorter J[6], Osman H[7]*
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336P Quantifying the burden-of-illness of Duchenne Muscular Dystrophy in Belgium: an interim analysis of a site-based survey

***De Waele L**[1,2], Cremers J[3], Debien E[3], Gielis E[1], Maenen V[1], Vanoppen I[3], Van Stappen T[4], Beeckman L[4], Posner N[5], Dukacz S[5], Jiang L[5], Evans J[6], Wu Y[6], Jones C[6], Meeus L[7], Claeys K[3,8]*
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337P Functional assessments used in routine care for Duchenne Muscular Dystrophy in the USA: a survey of Certified Duchenne Care Centers

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338P Very late-onset Becker Muscular Dystrophy caused by a new DMD variant

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339P Exploring the content validity of Patient-Reported Outcome (PRO) measures to capture the patient experience of Becker Muscular Dystrophy (BMD)

***Bronson A**[1], Mellor K[2], Kilburn N[1], Marshall C[1], Al-zubeidi T[2]*
[1]Edgewise Therapeutics, [2]Clarivate

340P Upper extremity assessment in Becker Muscular Dystrophy: a functional and MRI data approach

***Bellam P**[1], Rock K[1], Willcocks R[1], Barnard A[1], Forbes S[1], Lott D[1], Senesac C[1], Rooney W[2], Baetscher E[2], Subramony S[1], Chahin N[1], Dixon L[1], Walter G[1], Vandenborne K[1]*
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341P Incidental DMD in-frame deletions identified by prenatal array CGH: inter and intra-familial phenotype variability

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342P Use of psychopharmacological drug in Duchenne Muscular Dystrophy: a European multi-centre study

Muntoni F[1,11], *Weerkamp P*[2,3], *Miranda R*[4], *Collin P*[5], *Vroom E*[6], *Niks E*[7,13], *Vissing J*[8], *Desquerre I*[9], *Straub V*[10], *Mercuri E*[12], *Hendriksen J*[2,3,13]

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343VP Duchenne Muscular Dystrophy: the critical role of pulmonary infection in survival outcomes

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344P DMD-YOUNG: paediatric palliative care for children with a slowly progressive neuromuscular disorder in transition to adulthood

Annexstad E[1], *Ramberg C*[2], *Rosenberger A*[2], *Nordstrøm M*[1,3]

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345P Becker Muscular Dystrophy (BMD): natural history

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346P Cognitive profile of children with Duchenne and Becker Muscular Dystrophies

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347P Project HERCULES – building an evidence base to inform clinical care and access to medicines in DMD

Godfrey J[1], *Reuben E*[2], *Chandler F*[3]

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348P A national plan of action to raise Awareness and Improve Medical care of Duchenne Muscular Dystrophy (AIM-DMD)

Akinci G[1], *Coskun A*[2], *Koken O*[3], *Ardicli D*[4], *Cinar E*[1], *Okur T*[1], *Ayanoglu C*[5], *Bektas Ontas H*[4], *Ontas E*[6], *Cakir T*[7], *Komur M*[8], *Yuksel D*[9], *Yis U*[10], *Topaloglu H*[5]

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349P Comparison of short- and long-term proteomic response to the fast skeletal myosin inhibitor, sevasemten (EDG-5506), in Becker Muscular Dystrophy (BMD)

Barthel B[1], *Madden M*[1], *Thaler L*[1], *Evanchik M*[1], *Koch K*[1], *Donovan J*[1], *Collins S*[1], *Phan H*[2], ***Russell A***1

[1]*Edgewise Therapeutics,* [2]*Rare Disease Research*

350P Multi-parametric MRI of lower leg muscle in patients with Becker Muscular Dystrophy

Gerhalter T[1], *Schunk V*[1], *Baudin P*[2], *Rauh S*[3], *Tkotz K*[1], *Zaiss M*[1], *Roemer F*[1], *Dörfler A*[1], *Uder M*[1], *Gazzerro E*[4], *Nagel A*[1]

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351P Sevasemten, a fast myosin inhibitor, in adults with Becker Muscular Dystrophy results in reduced muscle damage biomarkers and functional stabilization

Donovan J[1], *Phan H*[2], *Russell A*[1], *Barthel B*[1], *Thaler L*[1], *Kilburn N*[1], *Amato M*[1], *MacDougall J*[1]
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352P Severe gastrointestinal problems in Duchenne Muscular Dystrophy: a case series

Zielman- Blokhuis A[2], *Tytgat K*[3], *Groothuis J*[1], ***Houwen S***[1]
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353P Applying a decision-making framework for expanded access to Duchenne Muscular Dystrophy

Rouet J[1], *Duro-Ocana P*[1], *Lawrence C*[1], *Zanker S*[1]
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354P 3-Year outcomes in Colombian patients under 7 years old with Duchenne Muscular Dystrophy early treated with ataluren

Toro C[1], *Ruiz-Ospina E*[2], *Castellar-Leones S*[3], *Ladino-Cortés L*[2], *Bolaños-Almeida C*[2], *Bobadilla-Quesada E*[2], *Maradei-Anaya S*[2], *Ortiz-Giraldo B*[4], *Acosta-Aragón M*[5], *Guerra-Araujo V*[11], *Alvarez-Montañez A*[6], *Silvera-Redondo C*[7], *Pachajoa H*[8], *Londoño-Mesa I*[9], *Torres-Nieto M*[11], *Del Río-Ospina L*[10]
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355P A comprehensive education and training programme for healthcare professionals in care and research for Duchenne Muscular Dystrophy developed from DMD Care UK and the DMD Hub

Turner C[1], *Geuens S*[2], *Heslop E*[1], *Johnson A*[3], *Straub V*[1], *Guglieri M*[1]
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356P A retrospective review of adult Duchenne Muscular Dystrophy patients in a UK centre of excellence and report the results of adherence to the standards of care guidelines for cardiac management

Willis T[1,2], *Kulshrestha R*[1], *Jones J*[1]
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357P Correlation between height, weight, and body mass index z-scores and clinical outcome assessments in young boys with Duchenne Muscular Dystrophy

Schiava M[1], *Dang U*[2], *Wood C*[3], *Wong S*[4], *Muni Lofra R*[1], *Ward L*[5], *Mayhew A*[1], *Tawil R*[6], *Willis T*[7], *Griggs R*[6], *Guglieri M*[1]
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358P Long-term changes of motor function in Becker Muscular Dystrophy

Bello L[1], *Riguzzi P*[1], *Capece G*[1], *Penzo M*[1], *Petrosino A*[1], *Sogus E*[1], *Mastellaro S*[1], *Caroli M*[1], *Villa M*[1], *Sabbatini D*[1], *Gorgoglione D*[1], *Vianello S*[1], *Sorarù G*[1], *Pegoraro E*[1]
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359VP Social difficulties and care burden of adult Duchenne Muscular Dystrophy in Japan: A questionnaire survey based on the Japanese Registry of Muscular Dystrophy (Remudy)

Mori-Yoshimura M[1], *Ishigaki K*[2], *Shimizu-Motohashi Y*[3], *Ishihara N*[4], *Yoshida S*[5], *Unuma A*[1], *Nakamura H*[6]
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360VP Making DMD/BMD research data FAIR: the case of heterogeneous brain (BIND) data

Siminiuc S[1], *Sakellariou E*[1], *Queralt Rosinach N*[3], *Pedomo Quinteiro P*[4], *Spitali P*[3], *Paliouras G*[1,2], *Muntoni F*[5]
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516P-536P, 537VP: Metabolic and mitochondrial myopathies

516P Fanconi Syndrome-induced severe Osteomalacic Myopathy: an uncommon side effect of Tenofovir Disoproxil Fumarate

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517P Clinical characteristics and genetic analysis of Chronic Progressive External Ophthalmoplegia

Choi S[1], *Lee S*[1], *Choi K*[1]
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518P Proteomic characterization of polyglucosan bodies in skeletal muscle in glycogenin-1 deficiency

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519P Insights into Primary Mitochondrial Myopathies: baseline characteristics and potential biomarkers from a natural history study

Martín Jiménez P[1], *Bermejo Guerrero L*[1], *Blázquez A*[2], *Serrano Lorenzo P*[2], *Hernández Lain A*[3], *Lucas Gómez B*[4], *Martín M*[5], *Domínguez González C*[1]
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520P Periodic paralysis in a child with Thermosensitive Mitochondrial Trifunctional Protein Deficiency

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521P Methodology and design of a study to characterize the disease course in a large dataset of patients with Thymidine Kinase 2 Deficiency

Domínguez-González C[1,2,3], *Nascimento A*[3,4], *Ma Y*[5], *Panahloo Z*[5], *Boudiaf N*[5], *Kim R*[6], *VanMeter S*[6], *Brunnert M*[7], *Hirano M*[8]
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522P Idiopathic hyperCKemia and myalgia- keep looking till you find a cause

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523P Rare Disease Register: McArdle Disease and Glycogenoses

Finnigan P[1], *Godfrey R*[1,2], *Arikan E*[1], *Bythel M*[3], *Aston J*[3], *Mohamed K*[1], *Quinlivan R*[1]
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524P Grip strength in McArdle disease

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525P Management of seizures in patients with Primary Mitochondrial Diseases: consensus statement from the inter-ERNs mitochondrial working group

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526P Clinical spectrum and treatment in paediatric *TANGO2* deficiency disorder

***Ortez Gonzalez C**[1], Ortuño B[1], Sarquella-Brugada G[1], Meavilla S[1], García-Cazorla Á[1], Expósito-Escudero J[1], O' Callaghan M[1], Natera De-Benito D[1], Darling A[1], Nascimento A[1], Julià N[1], Carrera-García L[1]*
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527P Small molecule adenylosuccinic acid supports muscle metabolism in *Adss1* deficient muscle phenotypes

***Rybalka E**[1,2,3], Kourakis S[1,2], Yates T[1], Bagaric R[1,2], Qi B[1,2], Ali B[1,2], Khandari N[4], Peterson A[5], Yan X[1], Kuang J[1], Nijagal B[5], Deveson-Lucas D[4], Timpani C[1,2,3]*
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528P A novel homozygous variant of uncertain significance in *ETFDH* suspected of causing severe Metabolic Syndrome and Myopathy

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529P Paucisymptomatic late onset Pompe disease: lessons from the clinical, radiological and histopathological long-term follow-up of an untreated patient

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530P Biallelic missense variants in *C2orf69* cause mitochondrial dysfunction associated with early-onset neurodegeneration and autoinflammation

Oh R[1,2], *Maier M*[3], *Blaser S*[1,4], *Cameron J*[1,5], *Lafreniere A*[1,6], *Hawkins C*[1,6], *Reversade B*[3,7,8,9], *Yoon G*[1,10]
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531P The French Registry of Glycogen Storage Disease Type 3

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532P Clinical, pathological, and genetic characteristics of 27 Spanish patients with POLG-related disorders

Bermejo Guerrero L[1], *Restrepo-Vera J*[2,3,4], *Martín – Jiménez P*[1], *Blázquez A*[5], *Serrano-Lorenzo P*[5], *Navarro-Riquelme M*[5], *Hernández-Laín A*[6], *Juntas-Morales R*[2], *Martí R*[3,4], *Martín M*[7], *Domínguez – González C*[1,4]
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533P Long-term follow-up Deoxynucleoside therapy for Late Onset Thymidine Kinase 2 Deficiency patients

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534P Clinical and genetic characterization of Brazilian patients with TK2 deficiency

Moreno C[1], *Tácio Quadros Santos Monteiro Fonseca A*[1], *Cunha Artilheiro M*[1], *Macedo Serafim da Silva A*[1], *de Paula Estephan E*[1], *Abdo Paiva M1, Gontijo Camelo C*[1], *Santos Pessoa A, Paranhos Miranda Covalesk A, Tomaselli P*[1], *Scarpellini G*[1], *Gurgel-Giannetti J*[1], *Holanda Mendonça R*[1], *Maroco Cruzeiro M*[1], *Marques Júnior W*[1], *Ferreira da Rosa Sobreira C*[1], *Hirano M*[1], *Andres Nascimento A*[1], *Schlesinger D*[1], *Zanoteli E*[1]
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535P Cytochrome c oxidase (COX) deficiency due to a novel homozygous *COX6B1* pathogenic missense variant

Oldfors C[1], *Jennions E*[2], *Olsson-Engman M*[3], *Visuttijai K*[1], *Wiksell Å*[4], *Fluriach Dominguez N*[5], *Kollberg G*[1], *Oldfors A*[1]
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536P Biallelic variants in *BCS1L* cause a novel neuromuscular phenotype with motor neuronopathy

Orbach R[1], *Maio N*[2], *Donkervoort S*[1], *Foley A*[1], *Chao K*[3], *Lehky T*[4], *Silverstein S*[1], *Potticary A*[1], *Rouault T*[2], *Butterfield R*[5], *Bönnemann C*[1]
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537VP Evaluation of the Scottish rhabdomyolysis and metabolic myopathy gene panel highlights limitations of genetic test criteria and a postcode lottery in access to testing

He Z[1], *Longman C*[10], *Farrugia M*[5], *Fletcher E*[2], *Robb Y*[2], *McWilliam C*[3], *Ross A*[4], *Brennan K*[5], *Davenport R*[6], *Hopkins P*[7], *Horrocks I*[8], *Joseph S*[8], *Harvie J*[9], *Kerrigan S*[9], *Stewart K*[10], *Brennan K*[5], *Miller-Hodges E*[11]
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656P-676P: Pompe disease

656P AAV gene therapy in the Pompe canine model demonstrates correction of the muscle and CNS disorder with immune tolerance to human GAA

Byrne B[1], *Pope M*[1], *Gentry M*[1], *Sun R*[1], *Gurda B*[1], *Trivedi P*[1], *Cloutier D*[1], *Saba S*[1], *Fuller D*[1], *Coleman K*[1], *Corti M*[1]
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657P Cipaglucosidase alfa + miglustat in late-onset Pompe disease: two non-ambulatory patients switching from high-dose, high-frequency alglucosidase alfa

Byrne B[1], *Castelli J*[2], *Jain V*[2], *Sitaraman Das S*[2], *Zhang J*[2]
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658P High-risk screening for Late-Onset Pompe Disease in China: an expanded Multicenter Study

Zhu B[1,2,3], *Jiao K*[1,2,3], *Chang X*[4], *Yao X*[5], *Zhu W*[1,2,3]
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659P Unlocking long-term insights: antibody formation and efficacy of enzyme replacement therapy in adults with Pompe disease

Theunissen M[1], *Ditters I*[2], *van Doorn P*[1], *van den Hout J*[2], *Hoogeveen-Westerveld M*[3], *Jacobs E*[3], *van der Ploeg A*[2], *van der Beek N*[1]
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660P Safety and efficacy of switching to avalglucosidase alfa in patients with late-onset Pompe disease previously deteriorating on alglucosidase alfa

Potters L[1], *Theunissen M*[1], *Wagenmakers M*[2], *van Doorn P*[1], *van der Ploeg A*[3], *van der Beek N*[1]
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661P Sensitivity to Change and Meaningful Change Thresholds of the Quick Motor Function Test (QMFT) in Pompe Disease

van der Beek N[1], *Sjöström-Bujacz A*[2], *Daskalopoulou C*[3], *Papageorgiou D*[3], *An Haack K*[4], *Gallego V*[5], *DasMahapatra P*[5], *Thibault N*[5], *Zaher A*[6], *Armstrong N*[5], *Kruijshaar M*[1], *van der Ploeg A*[1]
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662P The novel *GAA* variant p.R190G: expanding the spectrum of late onset Pompe disease and possible implications for screening in the Chinese population

Jiao K[1], *Jacobs E*[2], *Yue D*[3], *Boonman J*[2], *Zhao C*[1], *Pijnappel W*[2], *Zhu W*[1]
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663P Screening for Pompe disease and its differential diagnoses

Sekulic A[1], *Todorovic T*[1], *Peric S*[1], *Basta I*[1], *Ivanovic V*[1], *Rakocevic-Stojanovic V*[1], *Bozovic I*[1], *Palibrk A*[1], *Viric V*[1]
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664P From past to present: Pompe disease, pseudodeficiency, and genetic challenges

Giliberto F[1,2], *Buonfiglio P*[3], *Luce L*[1,2,4], *Llames Massini C*[1,2], *Carcione M*[1,2]

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665P Spanish Pompe Registry: overview based on the 130 patients included

Martínez Marín R[1], *Reyes Leiva D*[2], *Nascimento Osorio A*[3], *Muelas N*[4], *Domínguez C*[5], *Rojas Marcos Í*[6], *Paradas C*[6], *Olivé M*[7], *Matas A*[8], *Gómez Caravaca M*[10], *Barba Romero M*[9], *Mendoza M*[11], *de León J*[12], *Gutiérrez A*[13], *Rabasa M*[14], *Segovia S*[7], *Díaz Manera J*[15]

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666P MyoScreen™ Infantile and Late-Onset Pompe human skeletal muscle disease models for drug discovery and development of next-generation gene therapies

Young J[1], *Morozzi G*[1], *Travard L*[1], *Yennek S*[2], *Lagerstedt J*[2], *Zablocki A*[1], *Martin A*[1], *Flaender M*[1], *Lorintiu O*[1], *Dupont A*[1], *Autier V*[1], ***Ventre E***[1], *Darimont B*[1], *Selig L*[1]

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667P Glycogen response to enzyme replacement therapy in patients with Pompe disease

Stemmerik M[1], *Vissing J*[1]

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668P Pompe Registry: Real-world experience of patients with late-onset Pompe disease who switched therapy from alglucosidase alfa to avalglucosidase alfa

Schoser B[1], *Toscano A*[2], *Foster M*[3], *Sparks S*[3], *Kishnani P*[4], *on behalf of the Pompe Registry sites*

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669P The FORCE(TM) platform enables TfR1-mediated delivery of enzyme replacement therapy to muscle and central nervous system, resolving Pompe pathology in mice

Picariello T[1], *Vieira B*[1], *Johnson J*[1], *Schlaefke L*[1], *Russo R*[1], *Chang A*[1], *Cui J*[1], *Yang S*[1], *Rinaldi S*[1], *Weeden T*[1], *Zanotti S*[1], *Beskrovnaya O*[1]

[1]*Dyne Therapeutics*

670P Post-baseline outcomes of the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in late-onset Pompe disease

Roberts M[1], *Cole D*[2], *Geberhiwot T*[3], *Hughes D*[4], *Lachmann R*[5], *Murphy E*[6], *Sharma R*[7], *Jain V*[8], *Moffat E*[9], *Rutecki J*[8], *Clarke S*[9], *Deegan P*[10]

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671P Miglustat: a first-in-class enzyme stabiliser for late-onset Pompe disease

Mozaffar T[1], *Roberts M*[2], *Byrne B*[3], *Dimachkie M*[4], *Hopkin R*[5], *Kishnani P*[6], *Schoser B*[7], *van der Ploeg A*[8], *Brudvig J*[9], *Fox B*[9], *Holdbrook F*[9], *Jain V*[9], *Johnson F*[9], *Zhang J*[9], *Parenti G*[10]

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672P New insights into Pompe disease using Raman-microscopy

Hintze S[1], *Krois E*[2], *Wieland K*[3], *Meinke P*[1], *Haisch C*[2], *Schoser B*[1]

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673P Magnetization transfer Imaging in Late-Onset Pompe disease

Croce M[1], Naz F[2], Barzaghi L[2,3,4], Paoletti M[3], Mongini T[5], Gasperini S[6], Filosto M[7], Maggi L[8], Sechi A[9], Grandis M[10,11], Sacchini M[12], Sciacco M[13], Vercelli L[5], Bonizzoni C[3], Bergsland N[14], Santini F[15,16], Deligianni X[15,16], Gandini in Wheeler-Kingshott C[1,17,18], Ravaglia S[1], Pichiecchio A[1,3]

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674P Quantification of skeletal muscle glycogen in Late Onset Pompe patients using carbon MR spectroscopy at 3 Tesla

Gonzalez Chamorro A[1,3], Wilson I[2], Bolaño C[1,3], Hollingsworth K[2], James M[1,3], Wong K[1,3], Fitzsimmons S[1,3], Diaz Manera J[1,3], Thelwall P[2]

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675P Understanding skeletal muscle pathology and function in Late-Onset Pompe disease: insights from Proteomic and Transcriptomic Analyses

Schaiter A[1], Lohanadan K[2], van der Ven P[2], Hentschel A[3], Mensch A[4], Hahn A[5], Kornblum C[6], Stenzel W[7], Krämer H[8], Rosenbohm A[9], Bartkuhn M[10], Roos A[11,12,13], Schänzer A[1]

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676P Impact of discontinuation and reintroduction of alglucoidase alpha in patients with late-onset Pompe disease

Andrejic N[1], Viric V[2], Andabaka M[3], Bozovic I[2], Palibrk A[2], Ivanovic V[2], Basta I[2], Stevic Z[2], Peric S[2]

[1]Medical Faculty University of Belgrade, [2]University Clinical Centre of Serbia - Neurology Clinic, [3]Clinic of Neurology and Psychiatry for Children and Youth, Belgrade

705LBP - 710LBP, 711VP, 712LBP - 717LBP, 719LBP - 733LBP,, 735LBVP, 736LP - 738LBP: Late Breaking

705LBP Epilepsy in childhood neuromuscular disorders: Is it truly rare?

Acar Arslan E[1], Öztürk G[1], İyişenyürek Ş[1], Akbeyaz H[1], Bıkmazer B[1], Karayakalı B[1], Ünver O[1], Türkdoğan D[1]

[1]Marmara University Pendik Research and Training Hospital

706LBP Evaluation of children diagnosed with muscular dystrophy panel

Ayca S[1], Orak S[1], Polat M[1], Çam S[2]

[1]Celal Bayar University, School of Medicine, Department of Pediatric Neurology, [2]Celal Bayar University, School of Medicine, Department of Medical Genetics

707LBP Transcriptomics reveals DMD-driven cell dynamics and mechanisms of fibroblast inflammatory tissue priming in human dystrophic muscle

Barthelemy F[1,2], Chesmore K[1,3], Scripture-Adams D[1,2], Nelson S[1,3,4,5], Miceli M[1,2]

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708LBP Clinical trial readiness in a rare and underserved disease: Learnings from community engagement and the lived experience in Becker muscular dystrophy (Becker)

Bronson A[1], *Cameron M*[3], *Tencer S*[1], *Engel P*[2], *Jackson S*[2], *Krieger K*[1], *Donovan J*[1]
[1]*Edgewise Therapeutics,* [2]*Engage Health,* [3]*AdvocacyWorks Consulting*

709LBP Longer-term efficacy, safety, and patient-reported outcomes of apitegromab in patients with nonambulatory SMA: Results from the 48-month TOPAZ study

Crawford T[1], *De Vivo D*[2], *Krueger J*[3], *Mazzone E*[4], *Song G*[5], *Marantz J*[5], *Gueye M*[5], *Yu D*[5], *Yao B*[5], *Umans K*[5], *Darras B*[6,7], *on behalf of the TOPAZ Study Team*[8]
[1]*John Hopkins Hospital, Department of Neurology,* [2]*Columbia University, Irving Medical Center,* [3]*Helen Devos Children's Hospital Neurology - Grand Rapids,* [4]*Catholic University,* [5]*Scholar Rock, Inc.,* [6]*Boston Children's Hospital, Department of Neurology,* [7]*Harvard Medical School,* [8]*TOPAZ Study Team includes clinical trial investigators, physical therapists, study coordinators, and Scholar Rock (sponsor) staff*

710LBP Exploring higher doses of nusinersen in spinal muscular atrophy: final results from part B of the 3-part DEVOTE study

Crawford T[1], *Finkel R*[2], *Mercuri E*[3], *Day J*[4], *Montes J*[5], *Garcia Romero M*[6], *Sumner C*[1], *Sohn J*[7], *Monine M*[7], *Paradis A*[7], *Sun P*[7], *Foster R*[8], *Gambino G*[8], *Makepeace C*[7], *Fradette S*[7], *Farewell R*[7], *on behalf of the DEVOTE Study Group*
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711LBVP Genetic susceptibility for thrombotic microangiopathies in a 10-month-old child affected by SMA type 1

D'Alessandro R[1,2], *Somà A*[1], *Salvalaggio A*[1], *Guarnone E*[1], *Cavallina I*[1], *Rolle E*[1], *Rossi F*[1], *Vacchetti M*[1], *Mongini T*[2], *Ricci F*[1]
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712LBP Epigenetic liquid biopsy for Duchenne and Becker muscular dystrophy

Hart S[1], *Fattal-Valevsky A*[2], *Lavi R*[2], *Sagi L*[2], *Shemer R*[1], *Dor Y*[1], ***Dor-Wolman T***[1]
[1]*Hadassh Hebrew Univesity Medical Center,* [2]*Tel Aviv Sourasky Medical Center*

713LBVP Heterozygous truncating variants in DAG1 are associated with sporadic and familial isolated hyperCKemia

Fiorillo C[1,7], *Traverso M*[1], *Baratto S*[1], *Panicucci C*[1], *Bruno C*[1,7], *Grandis M*[5,7], *Innes A*[3], *Torella A*[2,6], *Picillo E*[2,6], *Onore M*[2,6], *Politano L*[2,6], *Nigro V*[2,6], *Scala M*[1,7], *Barresi R*[4]
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714LBP Can 5q SMA neonatal screening be a sustainable reality in middle-income countries? The experience of Minas Gerais state in Brazil

Giannetti J[1], *Januário J*[1], *Milanez L*[1], *Sacramento A*[1], *Moura A*[1], *Braga T*[1], *Pinhati C*[1], *Carvalho N*[1]
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715LBP Juvenile ALS associated with the SPTLC1 gene: a diagnostic challenge

Gomez Montoya S[1], *Veneruzzo G*[2], *Monges S*[1]
[1]*Department of Neurology, Hospital de Pediatría J.P. Garrahan,* [2]*Genomics laboratory, Hospital de Pediatría J.P. Garrahan*

716LBP Distinct muscle regeneration capability of mesenchymal stromal cells derived from human iPS cells in Ullrich congenital muscular dystrophy model mice

Goto M[1], *Takenaka-Ninagwa N*[1], *Yoshioka-Bourgeois C*[1], *Miki M*[1], *Sakurai H*[1]
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717LBP PhenoRareAI: Phenotype-Based Intelligent Diagnosis for Rare Neuromuscular Disorders

Zhai W[1,2], ***Jiao K***[1], *Wang N*[1], *Yu L*[3], *Huang X*[4], *Li W*[5], *Hu C*[5], *Chen X*[1], *Zhu B*[1], *Zhang J*[1], *Chang X*[6], *Zhong H*[1], *Zhao C*[1], *Zhu S*[2], *Zhu W*[1]
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719LBP Duchenne muscular dystrophy gene editing therapy with CRISPR/high-fidelity Cas12Max

Luk A[1,4], *Lin J*[2], *Jin M*[2], *Li Z*[3], *Li T*[4], *Shi L*[4], *Yang H*[5], *Li G*[4]

[1]*HuidaGene Therapeutics,* [2]*Department of Neurology, First Affiliated Hospital, Fujian Medical University,* [3]*Lingang Laboratory,* [4]*HuidaGene (Shanghai) Therapeutics Co., Ltd,* [5]*Center for Excellence in Brain Science and Intelligence Technology, Chinese Academy of Sciences*

720LBP Initial data following 24 weeks of treatment with WVE-N531 in patients with DMD: findings from Part B (FORWARD-53) of a phase 1b/2 open-label study

Malhi S[1], *Servais L*[2], *Bader M*[3], *AlQurashi M*[4], *Tillinger M*[1], *Paulson D*[1], *Angelov A*[1], *Kaviya A*[1], *Hu X*[1], *Narayanan P*[1], *Hart A*[1], *Haegele J*[1], *Singh K*[1], *Rheinhardt J*[1], *Ghosh A*[1], *Saint S*[1], *Bhatia S*[1], *McClure T*[1], *Casey C*[1], *Li-Kwai-Cheung A*[1]

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721LBP Multi-modal benefits of deramiocel (CAP-1002) in late-stage patients with DMD: A new treatment approach to target skeletal and cardiac muscle pathogenesis (HOPE-2-OLE Trial – 36-month data)

McDonald C[1], *Villa C*[2], *Tian C*[2], *Zygmunt A*[2], *Soslow J*[3], *Ollberding N*[2], *Hor K*[4], *Harmelink M*[5], *Varadhachary A*[6], *Apkon S*[7], *Romana V*[8], *Desai U*[8], *Hogan N*[8], *Maharry K*[8], *Awadalla M*[8], *Marban L*[8]

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722LBP Evaluating the effectiveness of clinical tools in determining the causes of recurrent rhabdomyolysis

Nandakumar D[1], *Breland C*[1], *Bhai S*[1]

[1]*University Of Texas Southwestern Medical School*

723LBP The genetic diagnosis of neuromuscular diseases beyond whole genome sequencing: a next step in testing

Nigro V[1,2], *Di Letto P*[1], *Rahman S*[1], *Zanobio M*[1], *Romano F*[1], *Scarpato M*[1], *Vicidomini G*[1], *Giacobbe C*[1], *Picillo E*[1], *Spampanato C*[1,2], *Del Vecchio Blanco F*[1], *Piluso G*[1], *Politano L*[1], *Torella A*[1,2]

[1]*University of Campania Luigi Vanvitelli,* [2]*Telethon Institute of Genetics and Medicine*

724LBP The natural history of pediatric Becker muscular dystrophy: a single center retrospective cohort study and muscle ultrasound characterization

Piccoli C[1], *Glanzman A*[2], *Gross B*[2], *Yum S*[2]

[1]*Stanford University,* [2]*Children's Hospital of Philadelphia*

725LBP Meaningful Within-Patient Change (MWPC) thresholds for North Star Ambulatory Assessment (NSAA) and Modified Pediatric Outcomes Data Collection Instrument (PODCI) in patients with Duchenne Muscular Dystrophy (DMD)

Posner N[1], *Bushmakin A*[1], *Mazar I*[1], *Shen Q*[1], *Duckacz S*[1], *Ines M*[1], *Aslam Z*[1], *Cappelleri J*[1]

[1]*Pfizer, Inc.*

726LBP Long-term safety and tolerability of delandistrogene moxeparvovec in Duchenne muscular dystrophy: phase 1 to phase 3 clinical trials

Proud C[1], *McDonald C*[2], *Mercuri E*[3], *Muntoni F*[4], *Zaidman C*[5], *Dharia S*[6], *Mason S*[6], *Meng J*[6], *Murphy A*[7], *Palatinsky E*[6], *Reid C*[7], *Wandel C*[8], *Mendell J*[6]

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727LBP Evidence for a role of eotaxin family proteins as inflammatory mediators and biomarkers in inclusion body myositis: new insights in the disease pathogenesis

Riolo G[1], *Gibertini S*[1], *Iannibelli E*[1], *Carnazzi A*[1,2], *Nicolini De Gaetano L*[1], *Salerno F*[1], *Ruggieri A*[1], *Bonanno S*[1], *Marcuzzo S*[1], *Cavalcante P*[1], *Maggi L*[1]

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728LBP Post-Authorisation Safety Study (PASS). Mexiletine effectiveness in patients with non-dystrophic myotonia: interim analysis findings

Rosenbohm A[1], *Vicart S*[2], *Tard C*[3], *Jayaseelan D*[4], *Sedehizadeh S*[5], *Schneider-Gold C*[6], *Adetoro N*[7], *Zozulya-Weidenfeller A*[8]

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729LBP Protein biomarkers of muscle injury exhibit differential reduction with subject age and time in adults with Becker muscular dystrophy

Russell A[1], Barthel B[1], Schrama E[2], Koeks Z[2], van de Velde N[2], Verschuuren J[2], Donovan J[1], Niks E[2], Spitali P[2]
[1]Edgewise Therapeutics, [2]University of Leiden

730LBP Double trouble: a comprehensive study into unrelated genetic comorbidities in adult patients with Facioscapulohumeral muscular dystrophy type I

Saconni S[1,2], Tammam G[3,4], Ezaru A[1], Slioui A[1], Monforte M[5], Tasca G[6], Villa L[1], Cavalli M[1], Salviati L[7], van der Vliet P[8], Lemmers R[8], Pini J[1,2], van der Maarel S[8], Puma A[1]
[1]Peripheral Nervous System & Muscle Department, Pasteur 2 Hospital, Nice University Hospital, [2]Institute for Research on Cancer and Aging of Nice, CNRS, INSERM, Côte d'Azur University, [3]Department of Brain and Behavioral Sciences, University of Pavia, [4]IRCCS Mondino Foundation, [5]Institute of Neurology, Catholic University School of Medicine, [6]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, [7]Clinical Genetics Unit, Department of Women's and Children's Health, University of Padua, [8]Department of Human Genetics, Leiden University Medical Center

731LBP Efficient exon skipping and dystrophin restoration after a single in vivo dose of Antisense Oligonucleotide Conjugate (AOC) in the mdx mouse model for Duchenne muscular dystrophy

Stan T[1], Van De Vijver D[1], Tanganyika-de Winter C[1], Post E[2], van Geel R[2], Türkmen N[2], van Delft F[2], Aartsma-Rus A[1]
[1]Leiden University Medical Center (LUMC), [2]Synaffix BV

732LBP Post-exercise biomarkers of muscle injury are reduced by Sevasemten, a fast myosin inhibitor, in adults with Becker muscular dystrophy

Stemmerik M[1], Russel A[2], Frølich S[1], Receveur N[1], Slipsager A[1], Barthel B[2], Donovan J[2], MacDougall J[2], Vissing J[1]
[1]Copenhagen Neuromuscular Center, Rigshospitalet, [2]Edgewise Therapeutics

733LBP Development of KT323 as a potent and safe gene therapy candidate for facioscapulohumeral muscular dystrophy (FSHD) through high throughput AAV capsid and cargo engineering

Tabebordbar S[1], Seshadri S[1], Brown K[1], Dang C[1], Tabebordbar S[1], Marsh E[1], Salmun G[1], Hakim C[1], Jenquin J[1], Stricker J[1], Ferguson B[1], Fielden M[1]
[1]Kate Therapeutics

735LBP First in class ASO targeting IGHMBP2 cryptic splice variant: efficacy and safety

Tyner C[1], Smieszek S[1], Przychodzen B[1], Johnson C[1], Polymeropoulos C[1], Birznieks G[1], Polymeropoulos M[1]
[1]Vanda Pharmaceuticals Inc.

736LBP Solving challenging titinopathy cases via multi-omics

Zhang Y[1], Xu L[1], Lei Y[2], Bönnemann C[3], Chan S[2], Javed A[1]
[1]School of Biomedical Sciences, LKS faculty of Medicine, The University of Hong Kong, [2]Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong Kong, [3]Neuromuscular and Neurogenetic Disorders of Childhood Section, Neurogenetics Branch, National Institute of Neurological Disorders and Stroke, National Institutes of Health

737LBP Full-length RNA sequencing in FSHD identifies novel isoforms and intergenic transcripts activated by DUX4

Zheng D[1], van den Heuvel A[1], Balog J[1], M. Willemsen I[1], Kloet S[1], J. Tapscott S[2,3], Mahfouz A[1,4], M. van der Maarel S[1]
[1]Leiden University Medical Center, [2]Fred Hutchinson Cancer Center, [3]University of Washington, [4]Delft University of Technology

738LBP Non-invasive optoacoustic muscle imaging in Late-onset Pompe disease – a translational and multicenter approach

Wagner A[1,2], Tan L[2,3], Zschüntzsch J[4], Meyer S[4], Stobbe A[4], Bruex H[4], Regensburger A[2,3], Alves F[4,5], Jüngert J[3], Rother U[6], Danko V[2,3], Türk M[7], Schmidt S[8], Vorgerd M[9,10], Schlaffke L[9], Wölfle J[3], Hahn A[11], Mensch A[12], Winterholler M[13], Trollmann R[3], Heiß R[8], Raming R[2,3], Knieling F[2,3]
[1]Department of Pediatric Neurology, Charité University Hospital Berlin, [2]Translational Pediatrics, Department of Pediatrics and Adolescent Medicine, University Hospital Erlangen, [3]Department of Pediatrics and Adolescent Medicine, University Hospital Erlangen, [4]Translational Molecular Imaging, Max-Planck Institute for Multidisciplinary Sciences (MPI-NAT), [5]Clinic for Haematology and Medical Oncology, Institute of Diagnostic and Interventional Radiology, University Medical Center Göttingen, [6]Department of Vascular Surgery, University Hospital Erlangen, [7]Department of Neurology, University Hospital Erlangen, [8]Institute of Radiology, University Hospital Erlangen, [9]Department of Neurology, BG-University Hospital Bergmannsheil; Ruhr-University Bochum, [10]Heimer Institute for Muscle Research; BG-University Hospital Bergmannsheil, [11]Department of Child Neurology; Justus-Liebig-Universität Giessen, [12]Department of Neurology; Martin-Luther-Universität Halle-Wittenberg, [13]Sana Krankenhaus Rummelsberg

16:45-17:15	**Short Oral Presentations 10** North Hall **181P, 177P, 178P, 180P, 179P** *Moderator: Laurent Servais, University of Oxford*	**Short Oral Presentations 11** Terrace 2A **656P, 675P, 673P, 674P, 358P** *Moderator: Pascal Laforêt, Hôpital Raymond Poincaré*	**Short Oral Presentations 12** Terrace 2B **536P, 109P, 513P, 512P, 241P** *Moderator: Heike Kölbel, Universitätsmedizin Essen*
19:15-01:00	**Networking Dinner** Municipal House (separate registration required)		

Saturday 12th October 2024

07:15-14:00	**Congress desk open**
07:15-14:00	**Myology Café open** Congress Hall Foyer
07:45-08:45	**Clinical Trial Updates** Congress Hall *Moderators: Francesco Muntoni, University College London, Great Ormond Street Hospital, UK & Tina Duong, Stanford University, USA*
07:45-08:00	**19O** Muscle MRI outcomes in patients with Duchenne Muscular Dystrophy treated with delandistrogene moxeparvovec: Findings from EMBARK Part 1 ***Vandenborne K***[1], *Walter G*[2], *Straub V*[3], *Willcocks R*[1], *Forbes S*[1], *Ennamuri S*[4], *Ding K*[4], *Reid C*[5], *Murphy A*[5], *Manfrini M*[6], *Elkins J*[4], *Rodino-Klapac L*[4] [1]*Department of Physical Therapy, University of Florida,* [2]*Department of Physiology and Aging, University of Florida,* [3]*Newcastle University, John Walton Muscular Dystrophy Research Centre,* [4]*Sarepta Therapeutics, Inc.,* [5]*Roche Products Ltd,* [6]*F. Hoffmann-La Roche Ltd*
08:00-08:15	**20O** Preliminary results from a Phase 1-2 gene therapy study of ATA-100, AAV9 vector encoding FKRP, in patients with Limb Girdle Muscular Dystrophy R9 ***Olivier S***[1], *Richard I*[1,2], *Stojkovic T*[3], *Straub V*[4], *Preisler N*[5], *Zanfongnon R*[2], *Buscara L*[2], *Genries-Ferrand S*[2], *Vissing J*[5] [1]*Atamyo Therapeutics,* [2]*Genethon,* [3]*APHP, Reference center for neuromuscular diseases, Institute of Myology,* [4]*John Walton Muscular Dystrophy Research Centre,* [5]*Rigshospitalet*
08:15-08:30	**22O** MExiletine versus lamotrigine in Non-Dystrophic myotonias – a randomised, double-blinded, cross-over trial ***Vivekanandam V***[1,2], *Jayaseelan D*[1], *Skorupinska I*[1], *Germain L*[1], *Matthews E*[3], *Barohn R*[4], *McDermott M*[5], *Hanna M*[1,2] [1]*The National Hospital for Neurology and Neurosurgery, Queen Square National Hospital for Neurology and Neurosurgery, Queen Square,* [2]*Queen Square UCL Institute of Neurology,* [3]*Neurosciences and Cell Biology Research Institute, St George's University of London,* [4]*University of Missouri,* [5]*University of Rochester Medical Center*
08:30-08:45	**21O** Rainbowfish: 2-year efficacy and safety data of risdiplam in infants with presymptomatic SMA ***Servais L***[1,2], *Finkel R*[3], *Farrar M*[4], *Vlodavets D*[5], *Zanoteli E*[6], *Al-Muhaizea M*[7], *Araújo A*[8], *Nelson L*[9], *Jaber B*[10], *Gorni K*[11], *Kletzl H*[12], *Palfreeman L*[13], *Gaki E*[13], *Rabbia M*[14], *Summers D*[13], *Fontoura P*[11], *Bertini E*[15], *on behalf of the RAINBOWFISH Study Group* [1]*MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford,* [2]*Division of Child Neurology, Centre de Références des Maladies Neuromusculaires, Department of Pediatrics, University Hospital Liège & University of Liège,* [3]*Center for Experimental Neurotherapeutics, St Jude Children's Research Hospital,* [4]*Sydney Children's Hospital Network and UNSW Medicine, UNSW Sydney,* [5]*Russian Children Neuromuscular Center, Veltischev Clinical Pediatrics and Pediatric Surgery Research Institute of Pirogov Russian National Research Medical University,* [6]*Department of Neurology, Faculdade de Medicina, Universidade de São Paulo,* [7]*Department of Neurosciences, King Faisal Specialist Hospital & Research Center-Riyadh,* [8]*Pediatrics Department, Faculty of Medicine, Federal University of Rio de Janeiro,* [9]*Department of Physical Therapy, University of Texas Southwestern Medical Center,* [10]*Pharma Development, Safety, F. Hoffmann-La Roche Ltd,* [11]*PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd,* [12]*Roche Pharmaceutical Research and Early Development, Roche Innovation Center Basel,* [13]*Roche Products Ltd,* [14]*Genentech, Inc.,* [15]*Research Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Research Hospital IRCCS*
08:45-09:00	**Comfort Break**

09:00-09:30	**Victor Dubowitz Lecture** Congress Hall *Moderators: Volker Straub, Newcastle University, UK & Jana Haberlová,University Hospital Motol, Czechia* **INV13** AAV delivery of mini-and full-length dystrophins ***Chamberlain J***[1], *Hauschka S*[1], *Regnier M*[1], *Tasfaout H*[1] [1]*University of Washington School of Medicine*
09:30-11:00	**Poster Highlights** *Moderators: Edoardo Malfatti, Université Paris Est, France & Tamara Dangouloff, University Of Liege, Belgium* **09:30- 09:45 23O** Spinal Muscular Atrophy is also a disorder of spermatogenesis ***Magot A***[1], *Reignier A*[2], *Binois O*[3], *Vuillerot C*[4,5], *Yann P*[1] *and the Fermasi Study Group* [1]*Centre de Référence des Mala Neuromusculaires AOC, CHU de Nantes, Filnemus, Euro-NMD, Nantes,* [2]*Service de Médecine et Biologie de la Reproduction, gynécologie médicale, CHU de Nantes,* [3]*Service de Biologie de la Reproduction - CECOS, Hôpital Antoine Béclère, AP-HP, Université Paris Saclay,* [4]*Centre de Référence PACA Réunion Rhône Alpes, Hospices Civils de Lyon, Hôpital Femme-Mère-Enfant, L'Escale, Service de Médecine Physique et de Réadaptation Pédiatrique, Bron,* [5]*NeuroMyogen Institute, CNRS UMR 5310 - INSERM U1217, University of Lyon* **09:45-10:00 24O** Temporal requirement of dystroglycan glycosylation during brain development and rescue of cortical dysplasia via gene delivery in the fetal stage ***Sudo A***[1], *Kanagawa M*[2], *Kobayashi K*[2], *Endo M*[3], *Minami Y*[3], *Aiba A*[4], *Toda T*[1] [1]*Department of Neurology, The University of Tokyo,* [2]*Division of Molecular Brain Science, Kobe University,* [3]*Division of Cell Physiology, Kobe University,* [4]*Laboratory of Animal Resources, The University of Tokyo* **10:00-10:15 25O** Development of a CRISPR/CasX 4q telomeric region ablation strategy for FSHD1 using an isogenic hiPSC line and a FSHD1 fibroblast cell line ***Lama C***[1], *de Graaf N*[1,2], *Bou Akar R*[1], *Danaus P*[3], *Suel-Petat L*[4], *Nectoux J*[3], *Authier F*[1,5], *Relaix F*[1], *Richard I*[4], *Malfatti E*[1,5] [1]*Université Paris Est Créteil, INSERM, IMRB - Hôpital Henri-Mondor,* [2]*The Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud university medical center,* [3]*Assistance Publique - Hôpitaux de Paris, APHP, Centre Universitaire Paris, Hôpital Cochin, Laboratoire de Génétique et Biologie Moléculaires,* [4]*Généthon, INSERM, Unité de recherche Integrare, UMR_S951 - Université Paris-Saclay, Université d'Évry,* [5]*Reference Center for Neuromuscular Disorders, APHP Henri Mondor University Hospital* **10:15-10:30 26O** Understanding the clinical heterogeneity in myotonic dystrophy type 1: identifying clinical phenotypes using unsupervised clustering ***la Fontaine L***[1], *Imkamp M*[1], *'t Hoen P*[2], *van As D*[2], *Smulders F*[2], *Bruijnes J*[1], *de Kok J*[1], *Faber C*[1], *Merkies I*[1], *van Kuijk S*[1] [1]*Maastricht University Medical Centre,* [2]*Radboud University Medical Centrels* **10:30-10:45 27O** Mass spectrometry as a technique for robust quantification of titin and other large muscle disease-associated proteins ***Smolnikov A***[1], *Padoani D*[1], *Jurczyluk J*[1], *Su Z*[1], *Hamey J*[1], *Wilkins M*[1], *Yuen M*[1,2,3,4], *Oates E*[1,5] [1]*School of Biotechnology and Biomolecular Sciences, Faculty of Science, University Of New South Wales,* [2]*Kids Neuroscience Centre, Kids Research Institute, The Children's Hospital at Westmead,* [3]*Functional Neuromics, Children's Medical Research Institute, The University of Sydney,* [4]*Faculty of Medicine and Health, The University of Sydney, 5Sydney Children's Hospital Network* **10:45-11:00 28O** Minimal detectable change of the Revised Hammersmith Scale in patients with Spinal Muscular Atrophy ***O'reilly E*** [1, 2],*Stimpson G*[2], *Rohwer A*[1,2], *Baranello G*[1,2,3], *Muntoni F*[1,2,3], *Scoto M*[1,2,3], *On behalf of SMA REACH UK*[2] [1]*Great Ormond Street Hospital for Children NHS Foundation Trust,* [2]*Dubowitz Neuromuscular Centre, Institute of Child Health, UCL,* [3]*UCL NIHR GOSH Biomedical Research Centre*
11:00-11:30	**Morning refreshments, exhibition and posters**
11:30-13:30	**Late Breaking News** Congress Hall *Moderators: Ana Topf, John Walton Muscular Dystrophy Research Centre, UK & Alan H. Beggs, Boston Children's Hospital, Harvard Medical School, USA*

11:30 - 11:45 01LBO **Biallelic variants affecting the DST-b isoform cause a severe congenital myopathy presenting with arthrogryposis, muscular hypotonia and dilated cardiomyopathy**

Jacob M[1], *Kölbel H*[2], *Munot P*[3], *Kopajtich R*[1,4], *Achleitner M*[5], *Hahn A*[6], *Schänzer A*[7], *Weis J*[8], *Sewry C*[3,9,10], *Phadke R*[3], *Sukenik-Halevy R*[11,12], *Maroofian R*[13], *Goméz-Andrés D*[14], *Wilson L*[15], *Schara-Schmidt U*[2], *Winkelmann J*[1,4,16], *Roos A*[2,17,18], *Mayr J*[5], *Distelmaier F*[19], *Wagner M*[1,4,20]

[1]*Institute of Human Genetics, Klinikum rechts der Isar, Technical University of Munich, School of Medicine and Health, Munich,* [2]*Department of Neuropediatrics and Neuromuscular Centre for Children and Adolescents, Center for Translational Neuro- and Behavioral Sciences, University Hospital Essen, Duisburg-Essen University,* [3]*Dubowitz Neuromuscular Centre, Great Ormond Street Hospital,* [4]*Institute of Neurogenomics, Helmholtz Munich,* [5]*University Children's Hospital, Salzburger Landeskliniken (SALK), Paracelsus Medical University,* [6]*Department of Child Neurology, Justus-Liebig-University Giessen,* [7]*Institute of Neuropathology, Justus Liebig University Giessen,* [8]*Institute of Neuropathology, RWTH Aachen University Hospital,* [9]*Wolfson Centre for Neuromuscular Disorders, Robert Jones and Agnes Hunt Orthopaedic Hospital,* [10]*Cellular Pathology, Salford Royal Hospital NHS Foundation Trust, Northern Care Alliance,* [11]*Genetic Institute, Meir Medical Center,* [12]*School of Medicine, Faculty of Medical and Health Sciences, Tel Aviv University,* [13]*Centre for Neuromuscular Diseases, UCL Queen Square Institute of Neurology,* [14]*Department of Pediatric Neurology, Vall d'Hebron University Hospital, Vall d'Hebron Hospital Campus,* [15]*Department of Clinical Genetics, Great Ormond Street Hospital,* [16]*Munich Cluster for Systems Neurology (SyNergy),* [17]*Department of Neurology, Heinrich Heine University Düsseldorf,* [18]*Division of Neurology, Department of Medicine, Children' s Hospital of Eastern Ontario Research Institute, The Ottawa Hospital, and Brain and Mind Research Institute, University of Ottawa,* [19]*Department of General Pediatrics, Neonatology, and Pediatric Cardiology, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University,* [20]*Division of Pediatric Neurology, Developmental Medicine and Social Pediatrics, Department of Pediatrics, Dr. von Hauner Children's Hospital, Ludwig-Maximilian University (LMU) Munich*

11:45 - 12:00 02LBO **Expanding the genetic and phenotypic landscape of replication factor C complex-related disorders: RFC4 deficiency is linked to a multisystemic disorder**

Saito Y[1], *Morimoto M*[2], *Ryu E*[3,4,5], *RFC4 study group, Toro C*[2], *Myung K*[3,6], *Nishino I*[1], *Malicdan M*[2,7]

[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry,* [2]*National Institutes of Health Undiagnosed Diseases Program, National Human Genome Research Institute, National Institutes of Health,* [3]*Center for Genomic Integrity, Institute for Basic Science,* [4]*Department of Biological Sciences, Ulsan National Institute of Science and Technology,* [5]*Department of Biological Chemistry and Molecular Pharmacology, Harvard Medical School,* [6]*Department of Biomedical Engineering, Ulsan National Institute of Science and Technology,* [7]*Human Biochemical Genetics Section, Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health*

12:00 - 12:15 03LBO **Decoy gene therapy for myotonic dystrophy**

Furling D[1], *Arandel L*[1], *Sureau A*[1], *Cordier A*[1], *Moulay G*[1], *Rouxel C*[1], *Klein A*[1]

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

12:15 - 12:30 04LBO **Interim clinical data summary: A phase 1b/2a open-label, dose escalation study to evaluate the safety and clinical activity of intramuscular doses of an AAV9-based gene therapy (BB-301) administered to subjects with oculopharyngeal muscular dystrophy (OPMD) with dysphagia**

Amin M[1], *Achlatis E*[1], *Balou S*[1], *Steele C*[2], *Peladeau-Pigeon M*[2], *Barrett E*[2], *Meng D*[2]

[1]*NYU Grossman School of Medicine,* [2]*Swallowing Rehabilitation Laboratory, University Health Network*

12:30- 12:45 05LBO **A phase 1 study of antisense oligonucleotide NS-035 in patients with Fukuyama congenital muscular dystrophy**

Fujino G[1], *Kitamura A*[1], *Takahashi A*[1], *Maeda M*[1], *Kubota A*[1], *Tokuyama Y*[2], *Wada I*[2], *Kobayashi K*[3], *Komaki H*[4], *Taniguchi-Ikeda M*[5], *Ishigaki K*[6], *Toda T*[1]

[1]*The University of Tokyo,* [2]*The University of Tokyo Hospital,* [3]*Kobe University,* [4]*National Center of Neurology and Psychiatry,* [5]*Fujita Health University Hospital,* [6]*Tokyo Women's Medical University*

12:45-13:00 06LBO **CIFFREO, a phase 3, randomized, double-blind, placebo-controlled study of fordadistrogene movaparvovec (FM) in ambulatory participants with Duchenne muscular dystrophy (DMD)**

Muntoni F[1], *Nascimento A*[2], *Shin J*[3], *Guglieri M*[4], *Stettner G*[5], *Veerapandiyan A*[6], *Gallo S*[7], *Shi H*[7], *Gundapaneni B*[7], *Neelakantan S*[7], *Lobello K*[7], *Shen Q*[7], *Levy D*[7], *Mercuri E*[8]

[1]*UCL GOS Institute of Child Health,* [2]*Passeig de Sant Joan de Déu,* [3]*Pusan National University School of Medicine,* [4]*Newcastle University,* [5]*University of Zürich,* [6]*University of Arkansas for Medical Sciences,* [7]*Pfizer Inc,* [8]*Universita' la Cattolica*

Prize Giving Ceremony *Moderator: Marco Savarese, University of Helsinki, Finland*

Introduction to the WMS 2025 Congress, Vienna, Austria

Handover of the WMS flag and close of Congress *Moderator: Volker Straub*

13:30-14:30 **Homeward lunch**