Saturday, July 7, 2018

17:15 - 18:30 **Guided Poster Session**
Room: Mezzanine Floor Gallery and Foyers

- Topic Group 1: Muscle Diseases of Genetic Origin and Acquired Myopathies: Clinical Features, Pathophysiology, Therapy

PS1Group1-001 **A FAMILY-BASED STUDY INTO PENETRANCE IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 1**
*M. Wohlgemuth*1, R.J. Lemmers2, M.A. Jonker3, E.L. Van Der Kooi4, C.G. Horlings1, B.G. Van Engelen1, S.M. Van Der Maarel5, G.W. Padberg6, N.C. Voermans1; 1Neurology/Nijmegen/NL, 2Human genetics/Leiden/NL, 3Health sciences/Nijmegen/NL, 4/Leeuwarden/NL, 5Human Genetics/Leiden/NL, 6Dep. Of Neurology/Nijmegen/NL

PS1Group1-002 **EVALUATING THE USEFULNESS OF NEW LINE IMMUNOASSAYS FOR MYOSITIS ANTIBODIES IN CLINICAL PRACTICE: A RETROSPECTIVE STUDY**
*F. Montagnese*1, H. Babacic1, P. Eichhorn2, B. Schoser3; 1Neurology/Munich/DE, 2Institute of Laboratory Medicine/Munich/DE, 3Friedrich-Baur Institute, Departement of Neurology/Munich/DE

PS1Group1-003 **EXPRESSION OF DP116 IS A PREDISPOSING FACTOR FOR CARDIAC . DYSFUNCTION IN DUCHENNE MUSCULAR DYSTROPHY**
*T. Yamamoto*1, A. Hiroyuki1, T. Imanishi1, Y. Nakamachi1, M. Matsuo2, K. Iijima3, J. Saegusa1; 1Department of Clinical Laboratory/--, Kusunoki-cho, Chuo-ku, Kobe/JP, 2Department of Physical Therapy, Faculty of Rehabilitation/Kobe/JP, 3Department of Pediatrics/Kobe/JP

PS1Group1-004 **RIMEPORIDE: RESULTS FROM A PHASE IB STUDY IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY**
T. Gidaro1, L. Servais1, S. Previtali2, A. Zambon2, J. Pitchforth3, K. Maresh3, J. Diaz-Manera4, P.G. Carlier5, P.Y. Baudin6, B. Marty5, S. Carnesecchi7, C. Laveille8, J. Gray9, *F. Porte Thome*9, D. Labolle9, M. Annousamy1, V. Chê1, M.G. Natali Sora2, S. Gerevini10, N. Vidal4, K. Groves11, F. Muntoni3; 1I-Motion/Paris/FR, 2Neurology/Milano/IT, 3/London/GB, 4Neuromuscular disorders Unit/Barcelona/ES, 5AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 6/Froyennes/BE, 7Department of Pathology and Immunology/Geneva/CH, 8/Lyon/FR, 9R&D/Plan Les Ouates/CH, 10Department of Neuroradiology/Milano/IT, 11Great Ormond Street Hospital for Children/London/GB

PS1Group1-005 **A LONG TERM PREVENTIVE EFFICACY STUDY WITH RIMEPORIDE, A SODIUM-PROTON EXCHANGER INHIBITOR, IN GRMD DOGS**
I. Barthélémy1, J. Su2, Y. Fromes3, S. Carnesecchi4, *F. Porte Thome*5, P.G. Carlier3, B. Ghaleb2, S. Blot1; 1U955 - IMRB, Inserm/Maisons-Alfort/FR, 2Inserm UMR 955/Créteil/FR, 3AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 4Department of Pathology and Immunology/Geneva/CH, 5R&D/Plan Les Ouates/CH

PS1Group1-006 **PREDICTORS OF EARLY LEFT VENTRICULAR SYSTOLIC DYSFUNCTION IN DMD PATIENTS**

R.H.D. Cirino1, R.H. Scola2, *R.D. Ducci*2, A.C.C. Wermelinger1, C.S.K. Kay2, P.J. Lorenzoni2, L.C. Werneck2, E.R. Carmes3, C.L.P. Da Cunha1; 1Cardiology/Curitiba/BR, 2Neurology/Curitiba/BR, 3Internal Medicine/Curitiba/BR

PS1Group1-007 **GENE THERAPY BY CRISPR-CAS9 MEDIATED EXON SKIPPING IN A PRE-CLINICAL MODEL OF DMD, THE GRMD DOG**

*I. Punzón*1, I. Barthélémy1, F. Auradé2, N. Blanchard-Gutton1, F. Piétri-Rouxel3, F. Relaix2, S. Blot1; 1Neurobiology/Maisons Alfort/FR, 2Henri Mondor Hospital School of Medicine/Créteil Cedex/FR, 3/Paris/FR

PS1Group1-008 **IN VIVO CELL TRACKING OF CANINE MYOBLASTS BY SODIUM/IODIDE SYMPORTER GENE EXPRESSION**

*I. Punzón*1, D. Mauduit1, I. Barthélémy1, N. Blanchard-Gutton1, J.-L. Thibaud2, P. De Fornel2, B. Holvoet3, J.-T. Vilquin4, M. Sampaolesi5, S. Blot1; 1ENVA, Neurobiology/Maisons Alfort/FR, 258 Rue Auguste Perret,/Créteil/FR, 3KU Leuven, Belgium/Leuven/BE, 4Myology Research Center, Pitié Salpêtrière Hospital,/Paris/FR, 5/Leuven/BE

PS1Group1-009 **CLINICAL OUTCOME STUDY OF DYSFERLINOPATHY: RELATIONSHIP BETWEEN MUSCLE MRI AND PHYSIOTHERAPY OUTCOME MEASURES**

*J.D. Manera*1, R. Fernández-Torrón2, 3, M. James2, A. Mayhew2, M. Eagle2, R. Muni Lofra2, F. Smith4, H. Sutherland2, A.M. Sawyer5, C. Tesi Rocha6, J.W. Day6, A. Peduto7, K.J. Jones8, E.M. Coppenrath9, M.C. Walter10, P.G. Carlier11, T. Stojkovic12, A. Blamire4, K. Bushby2, V. Straub13; 1Neuromuscular Unit, Neurology Department/Barcelona/ES, 2The John Walton Muscular Dystrophy Research Centre, Newcastle Hospitals Trust/Newcastle Upon Tyne/GB, 3Neuromuscular Area, Biodonostia Health Research Institute, Neurology Service, Donostia University Hospital/Donostia-San Sebastian/ES, 4Magnetic Resonance Centre, Institute for Cellular Medicine/Newcastle Upon Tyne/GB, 5Lucas Centre for Imaging/Stanford, CA/US, 6Department of Neurology and Neurological Sciences/Stanford, CA/US, 7Department of Radiology, Westmead Hosptial, Faculty of Health Sciences/Sydney/AU, 8Institute for Neuroscience and Muscle Research, Children's Hospital at Westmead/Sydney/AU, 9Department of Clinical Radiology/Munich/DE, 10Friedrich-Baur Institute, Departement of Neurology/Munich/DE, 11AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 12AP-HP, G.H. Pitié-Salpêtrière 47-83, Boulevard de l'Hôpital/Paris/FR, 13John Walton Muscular Dystrophy Research Centre/Newcastle Upon Tyne/GB

PS1Group1-010 **MUSCLE IMAGING AND CLINICAL OUTCOME MEASURES IN OCULOPHARYNGEAL MUSCULAR DYSTROPHY**

*H.M.J.M. Kroon*1, C.G. Horlings2, J.G. Kalf1, B.G. Van Engelen2; 1Rehabilitation/Nijmegen/NL, 2Neurology/Nijmegen/NL

PS1Group1-011 **DIGENIC SQSTM1-TIA1 MYOPATHY: CLINICAL AND PATHOLOGICAL FEATURES**

Z. Niu1, C.S. Pontifex2, S. Berini1, L.E. Hamilton2, E. Naddaf1, E. Wieben1, R.A. Aleff1, K. Martens2, A. Gruber3, A.G. Engel1, G. Pfeffer2, *M. Milone*4; 1/Rochester, MN/US, 2Clinical Neurosciences/Calgary, AB/CA, 3/Marshfield, WI/US, 4Neurology/Rochester, MN/US

PS1Group1-012 **PATHOLOGICAL AND GENETIC STUDIES IN PATIENTS WITH MENDELIAN DISORDERS OF MTDNA MAINTENANCE**
*D. Lehmann*1, A.E. Vincent2, H. Rosa2, M. Rocha2, S. Zierz3, R.W. Taylor2, D.M. Turnbull2; 1Department of Neurology/Ulm/DE, 2Institute of Neuroscience/Newcastle Upon Tyne/GB, 3Department of Neurology/Halle/S./DE

PS1Group1-013 **EPILEPSY CHARACTERIZATION IN LAMA2-RELATED CONGENITAL MUSCULAR DYSTROPHY**
*D. Natera De Benito*1, D. Itzep2, J. Muchart2, A. Ramirez2, D. Yubero2, L. Carrera2, J. Aparicio2, C. Jou2, C. Jimenez Mallebrera2, J. Colomer2, C. Ortez2, A. Nascimento2, V. San Antonio Arce2; 1Neuromuscular Disorders Unit/Barcelona/ES, 2/Barcelona/ES

PS1Group1-014 **A PHASE 1B/2 STUDY OF THE ANTI-MYOSTATIN ADNECTIN RG6206 (BMS-986089) IN AMBULATORY BOYS WITH DUCHENNE MUSCULAR DYSTROPHY**
K.R. Wagner1, B.L. Wong2, B. Byrne3, L. Sweeney3, L. Jacobsen4, G. Tirucherai4, M. Rabbia5, J. Buchbjerg6, D. Juergen6, M. Krishnan6, C. Bechtold4, *M. Castillo*6; 1/Baltimore, MD/US, 2Neurology/Cincinatti, OH/US, 3/Gainesville, FL/US, 4/Princeton, NJ/US, 5/New York, NY/US, 6/Basel/CH

PS1Group1-015 **IN VITRO FUNCTIONAL CHARACTERIZATION OF FKRP PATIENT MISSENSE MUTATIONS**
S.F. Dias Henriques, E. Gicquel, J. Marsolier, I. Richard; /Evry/FR

PS1Group1-016 **IDENTIFICATION OF MUTATIONS IN A COHORT OF UNCLASSIFIED INHERITED MUSCLE DISORDERS BY TARGETED NEXT GENERATION SEQUENCING**
*K. Polavarapu*1, A. Joshi2, V. Preethish-Kumar3, A. Mathur2, S. Nayak2, S. Ambawat2, S. Nashi4, S. Vengalil4, R. Santhosh5, G. Narayanappa5, M. Faruq2, A. Nalini6; 1Clinical Neurosciences, Neurology/Bangalore/IN, 2/New Delhi/IN, 3Clinical neurosciences, Neurology/Bangalore/IN, 4Neurology/Bangalore/IN, 5Neuropathology/Bangalore/IN, 6Neurology//IN

PS1Group1-017 **L-CITRULLINE AND METFORMIN DELAY MUSCLE DEGENERATION IN DUCHENNE MUSCULAR DYSTROPHY: RESULTS FROM A ANDOMISED CLINICAL TRIAL**
*P. Hafner*1, 2, U. Bonati1, 3, A. Klein1, 4, 5, D. Rubino1, V. Gocheva1, S. Schmidt1, 3, V. Laugel6, A. Capone7, M. Gloor8, O. Bieri8, T. Zumbrunn9, N. Gueven10, D. Fischer1, 2, 3; 1Division of Paediatric Neurology/Basel/CH, 2Division of Neurology/Bruderholz/CH, 3Department of Neurology/Basel/CH, 4Division of Paediatric Neurology/Berne/CH, 5Division of Paediatric Neurology/Lausanne/CH, 6/Strasbourg/FR, 7Division of Paediatric Neurology/Aarau/CH, 8Department of Radiology/Basel/CH, 9Department of Clinical Research/Basel/CH, 10Pharmacy, School of Medicine/Hobart/AU

PS1Group1-018 **MUTATION ANALYSIS IN MLPA NEGATIVE DUCHENNE MUSCULAR DYSTROPHY: NGS AS A DIAGNOSTIC TOOL PRIOR TO MUSCLE BIOPSY**
*K. Polavarapu*1, M.K. Saroja2, V. Preethish-Kumar3, D. Sekar4, S. Nashi5, S. Vengalil5, P.T. Thomas6, S.N. Rao2, A. Nalini7; 1Clinical Neurosciences, Neurology/Bangalore/IN, 2R&D Division/Bangalore/IN, 3Clinical neurosciences, Neurology/Bangalore/IN, 4/Bengaluru/IN, 5Neurology/Bangalore/IN,

6Department of Psychiatric Social Work/Bangalore/IN, 7Department of Neurology/Bangalore/IN

PS1Group1-019 **EZUTROMID SIGNIFICANTLY REDUCES MUSCLE DAMAGE IN DUCHENNE MUSCULAR DYSTROPHY**
F. Muntoni1, G. Layton2, I. Bhattacharya3, K. Vandenborne4, C. Faelan5, A. Heatherington3, D. Roblin2, R. Osahon2, *J. Tinsley*2, K. Davies6; 1/London/GB, 2/Abingdon/GB, 3/Cambridge, MA/US, 4/Gainsville, FL/US, 5/Westminster, CO/US, 6/Oxford/GB

PS1Group1-020 **NUMBER NEEDED TO TREAT IN SPINAL MUSCULAR ATROPHY TYPE 1 WITH AVXS-101 RELATIVE TO NUSINERSEN**
*O. Dabbous*1, M. Cloutier2, A. Guerin2, I. Pivneva2, E.Q. Wu2, D.M. Sproule3; 1/Bannockburn, AL/US, 2/Boston, MA/US, 3/Bannockburn, IL/US

PS1Group1-021 **A NOVEL NEXT-GENERATION THERAPY FOR POMPE DISEASE WITH IMPROVED EFFICACY IN MICE**
S. Xu, Y. Lun, R. Soska, A. Nair, M. Frascella, A. Garcia, A.S. Ponery, J. Feng, C. Della Valle, R. Gotschall, H. Do, K.J. Valenzano, R. Khanna; /Cranbury/US

PS1Group1-022 **THE MULTIPLE FACES OF ANTI-HMGCR ANTIBODY-RELATED MYOPATHIES**
P. Masrori, W. De Ridder, J. Baets; Neuromuscular Reference Centre, Departement of Neurology/Edegem/BE

PS1Group1-023 **A PLURIPOTENT STEM CELL-DERIVED MODEL OF MEROSIN DEFICIENT CONGENITAL MUSCULAR DYSTROPHY**
K. Lyon1, A. Rickard2, M. Hayhurst Bennett2, *U. Schmidt*2; 1College of Engineering/San Luis Obispo/US, 2/San Diego, CA/US

PS1Group1-024 **AN IN VITRO MODEL OF MYOTONIC DYSTROPHY TYPE 1 USING HUMAN EMBRYONIC STEM CELL-DERIVED SKELETAL MUSCLE**
E. Solana-Guizar1, S. Labarge2, M. Hayhurst Bennett2, *U. Schmidt*2; 1/San Marcos, CA/US, 2/San Diego, CA/US

PS1Group1-025 **DIMETHYLFUMARATE (DMF) DOWNMODULATES INFLAMMATION IN SKELETAL MUSCLE**
*K. Schmidt*1, S. Kaur1, A. Faust1, P. Balcarek2, J. Schmidt1; 1Department of Neurology/Göttingen/DE, 2Department of Trauma Surgery, Orthopaedics and Plastic Surgery/Göttingen/DE

PS1Group1-026 **TAMOXIFEN IN DMD: RATIONALE AND PROTOCOL FOR A MULTICENTRE, RANDOMISED, DOUBLE-BLIND, PLACEBO-CONTROLLED, PHASE 3 TRIAL**
*S. Schmidt*1, A.-L. Orsini1, P. Hafner1, D. Rubino-Nacht1, O.M. Dorchies2, A. Nascimento3, U. Schara4, S. Spinty5, H. Topaloglu6, D. Fischer1; 1/Basel/CH, 2School of Pharmaceutical Sciences/Geneva/CH, 3/Barcelona/ES, 4/Essen/DE, 5/Liverpool/GB, 6/Ankara/TR

PS1Group1-027 **MUSCLE HISTOPATHOLOGY IN INFANTILE DNM1L-RELATED MITOCHONDRIAL EPILEPTIC ENCEPHALOPATHY IS KEY FOR CLINICAL DIAGNOSIS**
*E. Bertini*1, D. Verrigni1, D. Battaglia2, L. Fusco3, L. Figà Talamanca4, R. Carrozzo1, D. Diodato1, A. D'Amico1, L. Papetti3, D. Ghezzi5, C. Lamperti5; 1Unit of Neuromuscular Disorders, Laboratory of Molecular Medicine/Rome/IT,

2Infantile Neuropsychiatric Unit/Rome/IT, 3Division of Neurology/Rome/IT, 4Division of Neuroradiology/Rome/IT, 5Division of Molecular Neurogenetics/Milano/IT

PS1Group1-028 **EFFECT OF A LONG-TERM TREATMENT WITH METFORMIN IN DYSTROPHIC MDX MICE: A RECONSIDERATION OF ITS THERAPEUTIC INTEREST IN DMD**

P. Mantuano1, R.F. Capogrosso1, F. Sanarica1, M.G. Morgese2, M. De Bellis1, A. Cozzoli1, A. Fonzino1, E. Conte1, G.M. Camerino1, L. Trabace2, *A. De Luca*1; 1Pharmacy - Drug Sciences/Bari/IT, 2Experimental and Clinical Medicine/Foggia/IT

PS1Group1-029 **IDENTIFICATION OF LATE-ONSET POMPE DISEASE AMONG HIGH-RISK POPULATION WITH NATIONWIDE SCREENING STUDY IN JAPAN**

*K. Ogata*1, M. Kosuga2, E. Takeshita3, T. Matsumura4, K. Ishigaki5, S. Ozasa6, H. Arahata7, K. Sugie8, T. Takahashi9, S. Kuru10, M. Kobayashi11, H. Takada12, A. Hattori13, M.P. Takahashi14, N. Tanaka15, T. Kimura16, M. Funato17, T. Okuyama18, H. Komaki19; 1Institute of Clinical Research / Department of Neurology/Hasuda, Saitama/JP, 2Center for Lysosomal Storage Diseases / Department of Clinical Laboratory Medicine / Division of Medical Genetics/Setagaya, Tokyo/JP, 3Department of Child Neurology/Kodaira, Tokyo/JP, 4Neurology/Toyonaka/JP, 5Department of Pediatrics/Shinjuku, Tokyo/JP, 6Department of Pediatrics/Kumamoto/JP, 7Department of Neurology/Omuta, Fukuoka/JP, 8Department of Neurology/Kashihara, Nara/JP, 9Department of Neurology/Sendai, Miyagi/JP, 10Department of Neurology/Suzuka, Mie/JP, 11Department of Neurology/Yurihonjo, Akita/JP, 12Department of Neurology/Aomori/JP, 13Department of Neonatology and Pediatrics/Nagoya, Aichi/JP, 14Department of Neurology/Suita, Osaka/JP, 15Department of Neurology/Yotsukaido, Chiba/JP, 16Department of Neurology/Asahikawa, Hokkaido/JP, 17Department of Pediatrics/Gifu/JP, 18Center for Lysosomal Storage Diseases / Department of Clinical Laboratory Medicine/Setagaya, Tokyo/JP, 19Department of Clinical Research Promotion / Department of Child Neurology/Kodaira, Tokyo/JP

PS1Group1-030 **CLINICAL COURSE OF ADULT POMPE DISEASE PATIENTS WHO DID NOT START OR DISCONTINUED ENZYME REPLACEMENT THERAPY**

*H.A. Van Kooten*1, L. Harlaar1, N.A.M.E. Van Der Beek1, P.A. Van Doorn1, A.T. Van Der Ploeg2, E. Brusse1; 1Department of Neurology, Center for Lysosomal and Metabolic Diseases/Rotterdam/NL, 2Department of Paedicatrics, Center for Lysosomal and Metabolic Diseases/Rotterdam/NL

PS1Group1-031 **PROLONGED EXERCISE TEST IN PATIENTS WITH HISTORY OF THYROTOXICOSIS**

C.-Y. Tan, H.-T. Tan, R.J. Ratnasingam, K.-J. Goh; /Kuala Lumpur/MY

PS1Group1-032 **CLINICAL AND HISTOPATHOLOGICAL FINDINGS IN MYOTONIC MUSCULAR DYSTROPHY TYPE 2: RETROSPECTIVE REVIEW OF 50 DNA-CONFIRMED CASES**

*B. Roy*1, Q. Wu2, C. Whitaker3, K. Felice3; 1Neurology/New Haven, CT/US, 2Pathology/Farmington, CT/US, 3Neurology/New Britain, CT/US

PS1Group1-033 **ASCERTAINMENT OF THE ADULT PATIENT COHORT WITH MITOCHONDRIAL DISEASE IN GLASGOW**

*M.E. Farrugia*1, C. Longman2, L. Snadden2, G.S. Gorman3, A.M. Schaefer3, R.W. Taylor3, Y.S. Ng3, D.M. Turnbull3, R. McFarland3, R.K. Petty1;

1Neurology/Glasgow/GB, 2Genetics/Glasgow/GB, 3Institute of Neuroscience/Newcastle Upon Tyne/GB

PS1Group1-034 **RESTING-STATE FMRI SHOWS ALTERED DEFAULT-MODE NETWORK FUNCTIONAL CONNECTIVITY IN DUCHENNE MUSCULAR DYSTROPHY PATIENTS**
*N. Doorenweerd*1, M. De Rover2, C. Marini-Bettolo3, K.G. Hollingsworth4, E.H. Niks5, J.G.M. Hendriksen6, H.E. Kan1, V. Straub7; 1C.J. Gorter Center for High Field MRI/Leiden/NL, 2Clinical psychology Unit/Leiden/NL, 3John Walton Muscular Dystrophy Research Center/Newcastle Upon Tyne/GB, 4Newcastle Magnetic Resonance Centre/Newcastle Upon Tyne/GB, 5Neurology/Leiden/NL, 6Neurological Learning Disabilities/Heeze/NL, 7John Walton Muscular Dystrophy Research Centre/Newcastle Upon Tyne/GB

PS1Group1-035 **SPECIFIC MUTATIONS IN MYBPC1 CAUSE MYOPATHY AND "MYOGENIC TREMOR"**
S. Jackson1, *J. Schaefer*1, A. Saak1, J. Stavusis2, B. Lace3, J. Geist4, I. Inashkina2, D. Pelnena2, S. Pajusalu5, N. Wright6, A. Kontrogianni-Konstantopoulos4, C.G. Bönnemann7; 1Neurology/Dresden/DE, 2/Riga/LV, 3/Quebec/CA, 4/Baltimore/US, 5/Tartu/EE, 6/Harrisonburg/US, 7/Bethesda, MD/US

PS1Group1-036 **DIAGNOSTIC APPROACH TO CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA – FROM CLINICAL EVALUATION TO GENETIC CONFIRMATION**
*B. Kierdaszuk*1, M. Kaliszewska2, K. Tonska2, E. Bartnik2, A.M. Kaminska1, A. Kostera-Pruszczyk1; 1Department of Neurology/Warsaw/PL, 2Institute of Genetics and Biotechnology, Faculty of Biology/Warsaw/PL

PS1Group1-037 **FEMALE FERTILITY IN MYOTONIC DYSTROPHY TYPE 1 AND 2**
*O. Parmova*1, I. Srotova1, M. Hulova1, E. Vlckova1, L. Mensova2, M. Podborska3, P. Stradalova1, E. Kralickova1, I. Crha4, R. Mazanec2, S. Vohanka1, J. Bednarik1; 1Neurology/Brno/CZ, 2Neurology/Prague/CZ, 3Clinical Biochemistry/Brno/CZ, 4Obstetrics and Gynecology/Brno/CZ

PS1Group1-038 **EFFECT OF AHK, A NOVEL MODULATOR OF RYANODINE RECEPTORS, IN DUCHENNE MUSCULAR DYSTROPHY**
*H. Lasa Fernandez*1, G. Aldanondo2, J. Lasa Elgarresta2, A. Irastorza3, J.I. Miranda3, J.M. Aizpurua3, A. López De Munain4, A. Vallejo Illarramendi2; 1Department of Neuroscience/Leioa/ES, 2Neuroscience Area/Donostia/ES, 3Faculty of Chemistry/Donostia/ES, 4Neurology/Donostia/ES

PS1Group1-039 **ASPIRO PHASE 1/2 GENE THERAPY TRIAL IN X-LINKED MYOTUBULAR MYOPATHY (XLMTM): PRELIMINARY SAFETY AND EFFICACY FINDINGS**
N. Kuntz1, P.B. Shieh2, B. Smith3, C.G. Bönnemann4, J.J. Dowling5, M.W. Lawlor6, W. Müller-Felber7, M. Noursalehi8, *S. Rico*8, L. Servais9, S. Prasad8; 1/Chicago, IL/US, 2/Los Angeles, CA/US, 3/Gainesville, FL/US, 4/Bethesda, MD/US, 5/Toronto, ON/CA, 6/Milwaukee, WI/US, 7/Munich/DE, 8/San Francisco, CA/US, 9/Paris/FR

PS1Group1-040 **MR IMAGING OF RESPIRATORY MUSCLE DYSFUNCTION IN POMPE DISEASE**
*L. Harlaar*1, P. Ciet2, A. Pittaro2, P.A. Wielopolski2, H.A. Van Kooten1, N.A.M.E. Van Der Beek1, E. Brusse1, A.T. Van Der Ploeg3, M. De Bruijne4,

H.A.W.M. Tiddens5, P.A. Van Doorn1; 1Department of Neurology, Center for Lysosomal and Metabolic Diseases/Rotterdam/NL, 2Department of Radiology and Nuclear Medicine/Rotterdam/NL, 3Department of Paediatrics, Division of Metabolic Diseases and Genetics, Center for Lysosomal and Metabolic Diseases/Rotterdam/NL, 4Biomedical Imaging Group Rotterdam, Departments of Radiology and Medical Informatics/Rotterdam/NL, 5Department of Paediatrics, Respiratory Medicine and Allergology, Department of Radiology and Nuclear Medicine/Rotterdam/NL

PS1Group1-041 **A CASE OF VLCAD DEFICIENCY MYOPATHY WITH NEW MUTATION AND FAVORABLE RESPONSE TO L-CARNITINE, RIBOFLAVIN, AND COQ10**
*F. Fatehi*1, Y. Nilipour2, S. Nafissi1; 1Neurology/Tehran/IR, 2Pathology/Tehran/IR

PS1Group1-042 **IMPAIRED INSULIN SIGNALLING IN SKELETAL MUSCLE OF MYOTONIC DYSTROPHY PATIENTS**
*L.V. Renna*1, F. Bose'1, E. Brigonzi2, G. Meola2, R. Cardani1; 1Laboratory of Muscle Histopathology and Molecular Biology/San Donato Milanese (mi)/IT, 2Department of Biomedical Sciences for Health/San Donato Milanese (mi)/IT

PS1Group1-043 **DISTAL MYOPATHIES WITH RIMMED VACUOLE IN IRAN, A CLINICAL, HISTOPATHOLOGICAL AND GENETIC REPORT OF A LARGE GROUP**
*Y. Nilipour*1, S. Nafissi2, F. Fatehi2, Y. Ashoorian1, N. Beladi Moghadam2, R. Boostani3, M. Rohani2, B. Haghi Ashtiani2, B. Zamani2, 4, K. Basiri5, F. Ashtari6, D. Fathi2, H. Shamshiri2; 1Pathology/Tehran/IR, 2Neurology/Tehran/IR, 3Neurology/Mashhad/IR, 4/Tehran/IR, 5/Isfahan/IR, 6Neurology/Isfahan/IR

PS1Group1-044 **GMPPB HOMOZYGOUS VARIANT IN ADULT ONSET LIMB GIRDLE MYASTHENIC SYNDROME: A LIKELY FOUNDER MUTATION**
*V. Preethish-Kumar*1, A. Töpf2, K. Polavarapu1, A. Joshi3, S. Balaraju4, A. Roos2, R. Horvath2, S. Nashi5, S. Vengalil6, A. Mathur3, S. Nayak3, S. Ambawat3, M. Faruq3, A. Nalini6, H. Lochmüller7; 1Clinical Neurosciences, Neurology/Bengaluru/IN, 2John Walton muscular dystrophy research center/Newcastle/GB, 3/New Delhi/IN, 4John Walton Muscular Dystrophy Research Center/Newcastle/GB, 5Neurology/Bangalore/IN, 6Neurology/Bengaluru/IN, 7Department of Neuropediatrics and Muscle Disorders/Freiburg/DE

PS1Group1-045 **NFAT5 AND P38 MAPKS INTERACT IN MUSCLE CELLS RESPONDING TO OSMOTIC AND INFLAMMATORY STRESS AND IN POLYMYOSITIS**
*B. De Paepe*1, S. Herbelet2, J. Schmidt3, J. De Bleecker4; 1Neuromuscular Reference Center/Ghent/BE, 2/Ghent/BE, 3Neurology/Goettingen/DE, 4Neurology/Ghent/BE

PS1Group1-046 **IMPLICATION OF THE BREAKPOINTS POSITION IN PATIENTS WITH THE MACRODELETION OF EXONS 45 TO 55**
J. Poyatos Garcia, C. Gomis Coloma, N. Muelas, P. Martí, J.J. Vílchez; Neurology/Valencia/ES

PS1Group1-047 **THE STUDY OF ALISKIREN IN MDX DYSTROPHIC MICE**

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PS1Group1-048 **DUCHENNE MUSCULAR DYSTROPHY: DO BOYS WITH A SHORTER STATURE MAINTAIN AMBULATION LONGER?**
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PS1Group1-049 **LOW LEVELS OF DYSTROPHIN PROTEIN ASSOCIATED WITH ATTENUATION OF DUCHENNE MUSCULAR DYSTROPHY PROGRESSION**
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PS1Group1-050 **TAMOXIFEN PROLONGS SURVIVAL, IMPROVES MOTOR FUNCTION AND REDUCES LEVELS OF DNM2 IN MTM1-NULL MICE, A MODEL OF XLCNM**
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PS1Group1-051 **ARE THERE DIFFERENT CLINICAL ENTITIES WITH DISTINCT DISEASE COURSE AMONG D4Z4 REDUCED ALLELE CARRIERS?**
*G. Ricci*1, F. Mele2, L. Ruggiero3, E. Bucci4, L. Maggi5, M. Govi2, F. Sera6, L. Vercelli7, L. Santoro3, T. Mongini7, L. Villa8, M. Moggio8, M. Filosto9, M. Scarlato10, S.C. Previtali10, M. Cao11, E. Pegoraro11, 12, R. Telese13, A. Di Muzio13, C. Rodolico14, G. Antonini4, M.G. D'Angelo15, A. Berardinelli16, R. Piras17, M.A. Maioli17, G. Tomelleri2, C. Angelini18, G. Siciliano1, R. Tupler2; 1Department of Clinical and Experimental Medicine/Pisa/IT, 2Department of Life Sciences/Modena/IT, 3Department of Neurosciences, Reproductive and Odontostomatological Sciences/Naples/IT, 4Department of Neuroscience, Mental Health and Sensory Organs, S. Andrea Hospital/Rome/IT, 5/Milan/IT, 6MRC Centre Epidemiology for Child Health/London/GB, 7Department of Neuroscience, Center for Neuromuscular Diseases/Turin/IT, 8Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Dino Ferrari Center/Milan/IT, 9Neurology Clinic, Spedali Civili Hospital/Brescia/IT, 10INSPE and Division of Neuroscience/Milan/IT, 11Department of Neurosciences/Padova/IT, 12/Padova/IT, 13Center for Neuromuscular Disease, CeSI/Chieti/IT, 14Department of Neurosciences, Policlinico "G. Martino",/Messina/IT, 15Department of Neurorehabilitation/Bosisio Parini/IT, 16Unit of Child Neurology and Psychiatry/Pavia/IT, 17/Cagliari/IT, 18/Venice/IT

PS1Group1-052 **A PH1/2 STUDY OF ENA® ANTISENSE OLIGONUCLEOTIDE (DS-5141B) WITH EXON 45 SKIPPING ACTIVITY IN PATIENTS WITH DMD**
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PS1Group1-053 **MOST LGMD2L MUTATIONS IN THE ANO5 GENE RESULT IN DECREASED PROTEIN LEVELS IN ANOCTAMINOPATHY PATIENTS**

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PS1Group1-054 **CRISPR/CAS GENE EDITING IN DUCHENNE MUSCULAR DYSTROPHY CULTURES TO TEST NEW TREATMENTS FOR THE DISEASE**
P. Soblechero-Martin, I. García-Jiménez, E. Ruiz-Del-Yerro, E. Albiasu-Arteta, V. Arechavala-Gomeza; Neuromuscular Disorders/Barakaldo/ES

PS1Group1-055 **EXTRA-SKELETAL MUSCLE MANIFESTATIONS OF FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY**
C. Kelly, E. Naddaf; /Rochester, MN/US

PS1Group1-056 **ANALYSIS OF CARDIAC TROPONIN T ALTERNATIVE SPLICING IN SKELETAL MUSCLE OF DM1 PATIENTS**
*F. Bose'*1, L.V. Renna2, N. Ferrari3, V. Labate4, M. Cavalli3, G. Meola5, R. Cardani6; 1Laboratory of Muscle Histopathology and molecular biology/San Donato Milanese (mi)/IT, 2Laboratory of Muscle Histopathology and Molecular Biology/San Donato Milanese (mi)/IT, 3Department of Biomedical Sciences for Health/Milan/IT, 4Department of Cardiology Heart Failure Unit/San Donato Milanese (mi)/IT, 5Department of Biomedical Sciences for Health/San Donato Milanese (mi)/IT, 6/San Donato Milanese (mi)/IT

PS1Group1-057 **IMPACT OF IDEBENONE ON RESPIRATORY BURDEN, INCLUDING RISK OF BRONCHOPULMONARY COMPLICATIONS, IN DUCHENNE MUSCULAR DYSTROPHY**
*O.H. Mayer*1, M. Leinonen2, S. Hasham2, T. Meier2, T. Voit3, G. Buyse For The Delos Study Group4; 1/Philadelphia, PA/US, 2/Pratteln/CH, 3/London/GB, 4/Leuven/BE

PS1Group1-058 **CLINICAL AND MOLECULAR FEATURES OF DISTAL MYOPATHIES IN A PORTUGUESE COHORT: REPORTING NOVEL GENE MUTATIONS**
C. Falcão De Campos, M. Oliveira Santos, I. Conceição, M. De Carvalho; /Lisbon/PT

PS1Group1-059 **DUCHENNE MUSCULAR DYSTROPHY AND THE HEART - HOW TO VISUALIZE BETTER? CASE SERIES REPORT**
R. Kalicanin Milanovic1, *A. Kosac*2, S. Djordjevic3, M. Bjelic3, M. Djukic3, J. Mladenovic2, J. Glumac2, T. Ristic2, M. Kiklic2, I. Jovanovic3, V. Milic Rasic2; 1/Novi Pazar/RS, 2Neurology/Belgrade/RS, 3/Belgrade/RS

PS1Group1-060 **RATIONALE FOR EDASALONEXENT DOSE SCHEDULE IN PHASE 2 OF THE MOVEDMD® TRIAL**
H. Liu1, R.S. Finkel2, H.L. Sweeney3, K. Vandenborne4, M. Mancini5, J.F. Reilly6, P. Bista6, D. Wachtel7, S. Chandran8, R. Dagher9, D. Picarella9, A. Fretzen8, A. Nichols10, *J. Donovan*5; 1DMPK and Clinical Pharmacology/Cambridge, MA/US, 2/Orlando, FL/US, 3Myology Institute/Gainesville, FL/US, 4Physical Therapy/Gainesville, FL/US, 5Clinical Operations/Cambridge, MA/US, 6Biology/Cambridge, MA/US, 7Biology Analytical/Cambridge, MA/US, 8Product Development/Cambridge, MA/US,

9Pharmacology & Toxicology/Cambridge, MA/US, 10Research and Development/Cambridge, MA/US

PS1Group1-061 **REPURPOSING TAMOXIFEN FOR SEVERE MYOPATHIES: FROM PRECLINICAL EVALUATION IN ANIMAL MODELS TO CLINICAL TRIALS IN PATIENTS**
*O.M. Dorchies*1, E. Gayi1, H.M. Ismail1, L.A. Neff1, X. Massana-Muñoz2, B.S. Cowling2, J.F. Laporte2, T. Dor3, D. Fischer4, U.T. Ruegg1, L. Scapozza1; 1School of Pharmaceutical Sciences/Geneva/CH, 2Department of Translational Medicine and Neurogenetics/Illkirch/FR, 3/Jerusalem/IL, 4/Basel/CH

PS1Group1-062 **THE CLINICAL VARIATION OF RYR1 GENE IN A LARGE FAMILY**
S. Jankelowitz; Central Clinical School/Sydney/AU

PS1Group1-063 **CLINICAL OUTCOME STUDY OF DYSFERLINOPATHY: WHAT ARE THE BEST FUNCTIONAL AND STRENGTH OUTCOME MEASURES FOR THIS POPULATION?**
*V. Straub*1, M. James1, A. Mayhew1, M. Jacobs2, 3, J. Feng2, S. Spuler4, K. Bushby1, J. Day5, K.J. Jones6, D.X. Bharucha-Goebel7, E. Salort-Campana8, A. Pestronk9, M.C. Walter10, C. Paradas11, T. Stojkovic12, M. Mori-Yoshimura13, E. Bravver14, J. Diaz-Manera15, E. Pegoraro16, J. Mendell17; 1John Walton Muscular Dystrophy Research Centre/Newcastle Upon Tyne/GB, 2Division of Biostatistic and Study Methodology, Children's National Health System/Dc, WA/US, 3Pediatrics, Epidemiology and Biostatistics/Washington Dc/US, 4Charite Muscle Research Unit/Berlin/DE, 5Department of Neurology/Palo Alto, CA/US, 6Institute for Neuroscience and Muscle Research, Children's Hospital at Westmead/Sydney/AU, 7Department of NeurolOGY/Washington Dc/US, 8Neuromuscular and ALS Center, La Timone Hospital/Marseille/FR, 9Department of Neurology/St. Louis, Mo/US, 10Dept. of Neurology, Ludwig-Maximilians-University/Munich/DE, 11Neuromuscular Unit, Department of Neurology/Sevilla/ES, 12AP-HP, G.H. Pitié-Salpêtrière 47-83, Boulevard de l'Hôpital/Paris/FR, 13Department of Neurology/Tokyo/JP, 14/Charlotte, Nc/US, 15Neuromuscular disorders Unit/Barcelona/ES, 16Department of Neuroscience/Padova/IT, 17Center for Gene Therapy/Columbus, OH/US

PS1Group1-064 **BREATHING IS LIFE! INSPIRATORY MUSCLE TRAINING IN CHILDREN AND ADOLESCENTS LIVING WITH NEUROMUSCULAR DISEASES**
*A. Human*1, J. Jelsma2, B.M. Morrow3; 1Physiotherapy/Garankuwa/ZA, 2Division Physiotherapy/Observatory/ZA, 3Paediatrics and Child Health/Rondebosch/ZA

PS1Group1-065 **PERSONALIZED MOLECULAR THERAPY IN A NEW TRANSLATIONAL LARGE ANIMAL MODEL FOR DUCHENNE MUSCULAR DYSTROPHY**
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PS1Group1-066 **DILATED CARDIOMYOPATHY AND LIMB-GIRDLE MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY REVEALING NEW DPM3 GENE MUTATIONS**

*J. Svahn*1, P. Laforet2, C. Vial1, N. Streichenberger3, N. Romero4, C. Bouchet-Séraphin5, N. Seta6, R. Menassa7, F. Bouhour1, G. Jousserand1, T. Stojkovic2; 1Electroneuromyography and Neuromuscular Department/Bron/FR, 2/Paris/FR, 3/Bron/FR, 4Unité de Pathologies neuromusculaires, Institut de Myologie/Paris/FR, 5Département de Biochimie et de Génétique/Paris/FR, 6Biochemistry Laboratory/Paris/FR, 7Department of Molecular Endocrinology and Rare Diseases/Bron/FR

PS1Group1-067 **DYSFERLINOPATHY: PHENOTYPIC ASPECTS IN 24 MOROCCAN PATIENTS AND BENEFITS FROM MUSCLE EXERCISE AND CORTICOIDS IN SOME CASES**
*N. Birouk*1, S. Sefiani2, B. Kably3, H. Belaïdi3, R. Ouazzani3; 1Clinical Neurophysiology Rabat/Rabat/MA, 2Neuropathology/Rabat/MA, 3Clinical Neurophysiology/Rabat/MA

PS1Group1-068 **LGMD DUE TO γ-SARCOGLYCAN DEFICIENCY (LGMD2C): STUDY OF NATURAL HISTORY IN 77 PATIENTS BELONGING TO 69 MOROCCAN FAMILIES**
*N. Birouk*1, A. Ghislain Mpandzou2, B. Kably3, S. Sefiani4, A. Sefiani5, H. Belaïdi3, R. Ouazzani3; 1Clinical Neurophysiology Rabat/Rabat/MA, 2Clinical neurophysiology Rabat/Rabat/MA, 3Clinical Neurophysiology/Rabat/MA, 4Neuropathology/Rabat/MA, 5Genetic/Rabat/MA

PS1Group1-069 **DOES A NORMAL DYSTROPHIN STAINING IN MUSCLE BIOPSY RULE1 OUT THE MOLECULAR DIAGNOSIS OF DUCHENNE / BECKER MUSCULAR DYSTROPHY?**
M. Ginzberg, T. Lerman-Sagie, D. Lev, R. Dabby, M. Sadeh, E. Leshinsky-Silver; /Holon/IL

PS1Group1-070 **ESYN- AND CK9- PROMOTORS DRIVE MUSCLE-SPECIFIC EXPRESSION OF TRANSGENES IN VITRO AND IN VIVO**
J. Meng, J. Counsell, S. Waddington, F. Muntoni, J. Morgan; /London/GB

PS1Group1-071 **E-CADHERIN IS ECTOPICALLY EXPRESSED IN THE MUSCLE FIBER OF INCLUSION BODY MYOSITIS**
C. Ikenaga, H. Ishiura, H. Date, A. Kubota, S. Tsuji, T. Toda, J. Goto, J. Shimizu; Department of Neurology/Tokyo/JP

PS1Group1-072 **CARE EVALUATION OF DUCHENNE MUSCULAR DYSTROPHY PATIENTS IN BRAZIL**
K.C. Turke, A.A.S. Carvalho, D. Feder; /Santo André/BR

PS1Group1-073 **GENOTYPE AND PHENOTYPE FEATURES OF BRAZILIAN PATIENTS WITH MCARDLE DISEASE**
A.A.S. Carvalho1, V.G. Silva2, M.E. Koch3, P.O. Delgado4, B.M. Bertassoli2, R. Corazzini4, *K.C. Turke*4, D.M. Christofolini5, I. Rodart5, F.L.A. Fonseca6, D. Feder2; 1Neurology/Santo Andre/BR, 2Pharmacology/Santo Andre/BR, 3/Santo Andre/BR, 4/Santo André/BR, 5Genetics/Santo Andre/BR, 6Laboratory Analysis/Santo Andre/BR

PS1Group1-074 **PROPOSED CUT-OFF FOR REACTIVITY OF ANTI-HMGCR AND ANTI-SRP ANTIBODIES IN PATIENTS STATIN-EXPOSED AND STATIN-UNEXPOSED**
A.A.S. Carvalho1, T.F. Vieira1, M.E. Koch1, V.G. Silva1, R. Corazzini1, P.O. Delgado1, *K.C. Turke*1, F.L.A. Fonseca2, D. Feder3; 1/Santo André/BR, 2Laboratory Analysis/Santo Andre/BR, 3Pharmacology/Santo Andre/BR

PS1Group1-075 **NECROTIZING MYOPATHY AFTER DENGUE: CASE REPORT**
A.A.S. Carvalho1, *K.C. Turke*1, D. Feder2; 1/Santo André/BR, 2Pharmacology/Santo Andre/BR

PS1Group1-076 **EXPRESSION OF DYSTROPHIN ISOFORMS IN NEW DUCHENNE MUSCULAR DYSTROPHY MICE MODEL**
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PS1Group1-077 **MYOTONIC DYSTROPHY TYPE 2 AS A MULTISYSTEM DISEASE**
I. Bozovic, *S. Peric*, J. Pesovic, B. Bjelica, M. Brkusanin, I. Basta, A. Marjanovic, M. Brankovic, A. Kacar, D. Savic-Pavicevic, V. Rakocevic-Stojanovic; /Belgrade/RS

PS1Group1-078 **HEART INVOLVEMENT IN MYOTONIC DYSTROPHY TYPE 2**
B. Bjelica, *S. Peric*, E. Cvijan, G. Mandic-Stojmenovic, M. Kovacevic, K. Aleksic, J. Pesovic, D. Savic-Pavicevic, I. Basta, D. Lavrnic, V. Rakocevic-Stojanovic; /Belgrade/RS

PS1Group1-079 **VARIANT REPEATS STABILIZE EXPANSION AND MODIFY AGE AT ONSET IN MYTONIC DYSTROPHY TYPE 1**
J. Pesovic, *S. Peric*, M. Brkusanin, G. Brajuskovic, V. Rakocevic-Stojanovic, D. Savic-Pavicevic; /Belgrade/RS

PS1Group1-080 **FEATURES OF THE SERBIAN COHORT OF PATIENTS WITH CALPAINOPATHY**
S. Peric, A. Kosac, M. Brankovic, A. Marjanovic, B. Banko, S. Milenkovic, M. Jankovic, D. Lavrnic, R. Maksimovic, V. Milic Rasic, V. Rakocevic-Stojanovic; /Belgrade/RS

PS1Group1-081 **MALIGNANT HYPERTHERMIA AND MH-LIKE REACTIONS IN NEUROMUSCULAR DISEASES**
B. De Wel1, *K.G. Claeys*2; 1Neurology/Leuven/BE, 2/Leuven/BE

PS1Group1-082 **ORO-PHARYNGEAL DYSPHAGIA IN MYOTONIC DYSTROPHY TYPE 1 (DM1): IDENTIFICATION OF SENSORY CHANGES IMPACTING SWALLOWING FUNCTION**
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PS1Group1-083 **HMGCR-MYOPATHY. A RARE BUT STILL A SEVERE DISEASE**
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PS1Group1-084 **VALUE OF DIAGNOSTIC ALGORITHM IN ASYMPTOMATIC HYPER-CK-EMIA AND ITERATIVE RHABDOMYOLYSIS**
*J.M. Grau*1, J.C. Milisenda1, F. Cardellach1, J. García2; 1Internal Medicine/Barcelona/ES, 2Biochemistry/Barcelona/ES

PS1Group1-085 **NEXT GENERATION SEQUENCING OF DYSTROPHIN GENE IN A COHORT OF NON-DELETION/DUPLICATION DMD/BMD EGYPTIAN PATIENTS**
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PS1Group1-086 **NATURAL HISTORY OF DISEASE-RELATED COMPLICATIONS IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY**
J. Lynch, K. Tsai, J. Lu, C. Mix, *A.M. York*; /Cambridge, MA/US

PS1Group1-087 **GOLODIRSEN LEADS TO SARCOLEMMAL DYSTROPHIN EXPRESSION IN PATIENTS WITH GENETIC MUTATIONS AMENABLE TO EXON 53 SKIPPING**
F. Muntoni1, D. Frank2, J. Morgan1, J. Domingos1, F. Schnell2, J.G. Dickson1, M. Guglieri3, A. Seferian4, M. Monforte5, L. Servais4, V. Straub3, E. Mercuri5, S.-N. Study Group2, *A.M. York*; 1/London/GB, 2/Cambridge, MA/US, 3/Newcastle Upon Tyne/GB, 4/Paris/FR, 5/Rome/IT

PS1Group1-088 **POMPE DISEASE IN FRANCE: MOLECULAR FEATURES, EPIDEMIOLOGY AND CLINICAL CORRELATIONS FROM A FOURTY-FIVE YEAR NATIONWIDE STUDY**
*C. Semplicini*1, P. Letard2, M. De Antonio3, N. Taouagh3, B. Perniconi3, F. Bouhour4, A. Echaniz-Laguna5, D. Orlikowski6, S. Sacconi7, E. Salort-Campana8, G. Solé9, F. Zagnoli10, D. Hamroun11, R. Froissart4, C. Caillaud3, P. Laforet6; 1/Padova/IT, 2/Paris, France/FR, 3/Paris/FR, 4/Lyon/FR, 5/Strasbourg/FR, 6/Garches/FR, 7/Nice/FR, 8/Marseille/FR, 9/Bordeaux/FR, 10/Brest/FR, 11/Montpellier/FR

PS1Group1-089 **A SPANISH MYOTONIC DYSTROPHY TYPE I FAMILY CARRYING INTERRUPTIONS SHOWING A MILDER AND ATYPICAL PHENOTYPE**
*E.A. Koehorst*1, 2, A. Ballester-Lopez1, 3, M. Almendrote1, A.R. Fransi1, 4, A. Martínez-Piñeiro1, 4, G. Lucente1, 4, I. Linares1, J. Nuñez1, N. Guanyabens5, A. Lucia6, A. Cano6, T. Casadevall7, A. Rodríguez-Palmero1, 8, L. Monlleó-Neila1, 8, G. Pintos-Morell1, 3, 8, J. Coll-Cantí1, 3, 8, G. Nogales-Gadea1, 3; 1Neuromuscular and Neuropaediatrics Group/Badalona/ES, 2Maastricht University/Maastricht/NL, 3Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER)/Madrid/ES, 4Neuromuscular Pathology Unit. Neurology Service. Neuroscience department/Barcelona/ES, 5Neurology Unit, Neuroscience department/Barcelona/ES, 6Universidad Europea/Madrid/ES, 7Neuromuscular Unit. Neurology Service. Neuroscience department/Barcelona/ES, 8Neuropediatric Unit. Pediatric Service./Barcelona/ES

PS1Group1-090 **MERRF CLASSIFICATION: IMPLICATIONS FOR DIAGNOSIS, AND CLINICAL TRIALS**
*J. Finsterer*1, S. Zarrouk-Mahjoub2, J.M. Shoffner3; 1/Vienna/AT, 2Pasteur Institute of Tunis/Tunis/TN, 3Molecular Genetics/Atlanta/US

PS1Group1-091 **MITOCHONDRIAL MULTI-ORGAN DISORDER SYNDROME SCORE GENERATED FROM DEFINITE MITOCHONDRIAL DISORDERS**
*J. Finsterer*1, S. Zarrouk-Mahjoub2; 1/Vienna/AT, 2Pasteur Institute of Tunis/Tunis/TN

PS1Group1-092 **CLINICAL PRACTICE WITH STEROID THERAPY FOR DUCHENNE MUSCULAR DYSTROPHY, A CLINICIAN SURVEY IN ASIAN AND OCEANIA**
*F. Takeuchi*1, H. Nakamura2, H. Komaki2, I. Nishino2, S. Takeda2, I. Nonaka2, N. Yonemoto3; 1The John Walton Muscular Dystrophy Research Centre/Newcastle Upon Tyne/GB, 2/Tokyo/JP, 3Department of Biostatistics/Kyoto/JP

PS1Group1-093 **EXPERIENCES WITH BARIATRIC SURGERY IN PATIENTS WITH FSHD AND DM1**
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PS1Group1-094 **AN UNUSUAL PRESENTATION OF GNE MYOPATHY WITH PROMINENT AXIAL MUSCLE WEAKNESS**
*J.-M. Park*1, J.-S. Park2; 1Neurology/Gyeongju/KR, 2Department of Neurology/Daegu/KR

PS1Group1-095 **HIGHER MRI MUSCLE FAT FRACTION AT SIMILAR AGE IS ASSOCIATED WITH EARLIER LOSS OF AMBULATION IN DUCHENNE MUSCULAR DYSTROPHY**
K.J. Naarding1, H. Reyngoudt2, E.W. Van Zwet3, M.T. Hooijmans1, B.L. Wong4, C. Tian4, I. Rybalsky4, K.C. Shellenbarger4, J. Le Louër2, P.G. Carlier2, H.E. Kan5, *E.H. Niks*1; 1Neurology/Leiden/NL, 2AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 3Department of Biostatistics/Leiden/NL, 4Neurology/Cincinatti, OH/US, 5C.J. Gorter Center for High Field MRI/Leiden/NL

PS1Group1-096 **TOR1A VARIANTS CAUSE A SEVERE ARTHROGRYPOSIS WITH DEVELOPMENTAL DELAY, STRABISMUS AND TREMOR**
A. Kariminejad1, M. Dahl-Halvarsson2, G. Ravenscroft3, F. Afroozan1, E. Keshavarz4, H. Goullée3, M. Davis, N. Laing, *H. Tajsharghi*; 1/Tehran/IR, 2Department of Pathology/Gothenburg/SE, 3Centre for Medical Research/Nedlands/AU, 4Department of Radiology/Tehran/IR

PS1Group1-097 **NDUFAF3 VARIANTS THAT DISRUPT MITOCHONDRIAL COMPLEX I ASSEMBLY CAUSE ASSOCIATE WITH CAVITATING LEUKOENCEPHALOPATHY**
*A. Ishiyama*1, K. Muramatsu2, S. Uchino3, 4, C. Sakai3, Y. Matsushima3, N. Makioka2, T. Ogata2, E. Suzuki2, H. Komaki1, M. Sasaki1, M. Mimaki3, 4, Y.-I. Goto3, I. Nishino5; 1Department of Child Neurology, National Center Hospital, National Center of Neurology and Psychiatry/Kodaira/JP, 2Department of Pediatrics, Gunma University Graduate School of Medicine/Maebashi/JP, 3Department of Mental Retardation and Birth Defect Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry/Kodaira/JP, 4Department of Pediatrics, Faculty of Medicine, Teikyo University/Itabashi/JP, 5Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry/Kodaira/JP

PS1Group1-098 **RESTRICTION ENZYME CLEAVAGE OF PCR PRODUCTS ALLOWS GENOTYPING MDX3CV, MDX4CV AND MDX5CV ALLELES WITHOUT SEQUENCING**
L.A. Neff, E. Gayi, L. Scapozza, O.M. Dorchies; School of Pharmaceutical Sciences/Geneva/CH

PS1Group1-099 **CLINICO-PATHOLOGICAL CORRELATIONS IN IDIOPATHIC INFLAMMATORY MYOPATHIES**

*E. Pál*1, K. Gulyas2, T. Minier2, T. Berki3, L. Czirjak2, C. Varju2; 1Neurology/Pécs/HU, 2Rheumatology and Immunology/Pécs/HU, 3Immunology and Biotechnology/Pécs/HU

PS1Group1-100 **FIRST REPORTED VARIANT AT THE TRIM32 RING DOMAIN IN A PEDIGREE WITH LIMB-GIRDLE MUSCULAR DYSTROPHY AND BARDET-BIEDL SYNDROME**
*A.L. Pelayo-Negro*1, M.A. Fernández-García2, P. Blanco-Arias2, E. Gallardo-Agromayor3, G. Gutierrez-Gutierrez4, J. Berciano1; 1Neurology/Santander/ES, 2Neurology/A Coruña/ES, 3Radiology/Santander/ES, 4Neurology/Madrid/ES

PS1Group1-101 **MUSCLE SPECIFIC KINASE PROTECTS MDX MOUSE MUSCLES AGAINST ECCENTRIC CONTRACTION-INDUCED LOSS OF STRENGTH**
W.D. Phillips, J. Ban, S. Trajanovska; Physiology & Bosch Institute/Sydney, NSW/AU

PS1Group1-102 **NOVEL LMNA GENE MUTATION PRESENTING WITH DILATED CARDIOMYOPATHY AND LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 1B**
M. Spilioti1, *K. Notas*1, G. Stavropoulos2, G. Efthimiadis3, K. Michaelidou4, I. Zaganas4, M. Moschou1, M. Arnaoutoglou1, M. Tsolaki1; 11st Department of Neurology, AHEPA General Hospital/Thessaloniki/GR, 22nd Cardiology Department, Hippokration General Hospital/Thessaloniki/GR, 31st Cardiology Department, AHEPA University Hospital/Thessaloniki/GR, 4Department of Neurology, School of Medicine/Heraklion, Crete/GR

PS1Group1-103 **LONGTERM APPLICATION OF HUMAN IMMUNOGLOBULIN G FOR EXPERIMENTAL TREATMENT OF DUCHENNE MUSCULAR DYSTROPHY**
*J. Zschüntzsch*1, P.V. Jouvenal1, Y. Zhang2, F. Klinker3, M. Tiburcy4, D. Liebetanz3, H. Brinkmeier2, D. Malzahn5, J. Schmidt1; 1Neurology/Göttingen/DE, 2Institute of Pathophysiology/Karlsburg/DE, 3Clinical Neurophysiology/Göttingen/DE, 4Institute of Pharmacology and Toxicology/Göttingen/DE, 5/Göttingen/DE

PS1Group1-104 **BODY COMPOSITION ANALYSIS IN PATIENTS WITH MYOTONIC DYSTROPHY TYPE 1 AND 2**
*S. Peric*1, M. Vujnic2, T. Nisic1, M. Banovic1, B. Bjelica1, I. Bozovic1, J. Pesovic1, D. Savic-Pavicevic1, I. Basta1, Z. Stevic1, D. Lavrnic1, V. Rakocevic-Stojanovic1; 1/Belgrade/RS, 2/Banja Luka/BA

PS1Group1-105 **EVALUATING THE EFFECTS OF BASELINE VARIABLES ON THE RESPIRATORY FUNCTION BENEFIT OF IDEBENONE IN DUCHENNE MUSCULAR DYSTROPHY**
*S. Hasham*1, T. Meier1, M. Leinonen1, T. Voit2, O.H. Mayer3, G. Buyse For The Delos Study Group4; 1/Pratteln/CH, 2/London/GB, 3/Philadelphia, PA/US, 4/Leuven/BE

PS1Group1-106 **NOVEL RYR-CALSTABIN STABILIZERS WITH THERAPEUTICAL POTENTIAL FOR DUCHENNE MUSCULAR DYSTROPHY**
G. Aldanondo, H. Lasa-Fernandez, J. Lasa-Elgarresta, J.I. Miranda, J.M. Aizpurua, A. López De Munain, *A. Vallejo-Illarramendi*; /Donostia-San Sebastian/ES

PS1Group1-107 **THE EFFECT OF UNCARIA TOMENTOSA IN DIAPHRAGM MUSCLE OF MDX DYSTROPHIC MICE**

D. Feder1, *A.A.S. Carvalho*2, B.M. Bertassoli1, L.P. Giordani1, G. Petri1, F.F. Perazzo3, B.C.A. Alves4, M.M. Perez4, F.L.A. Fonseca4; 1Pharmacology/Santo Andre/BR, 2Neurology/Santo Andre/BR, 3Pharmaceutical Sciences/Santo Andre/BR, 4Laboratory Analysis/Santo Andre/BR

PS1Group1-108 **MYTOBLOTS FOR THE EVALUATION OF NEW TREATMENTS IN NEUROMUSCULAR DISORDERS**

E. Ruiz-Del-Yerro1, P. Soblechero-Martin2, I. Larrañaga1, E. Albiasu-Arteta1, *V. Arechavala-Gomeza*1; 1Neuromuscular Disorders/Barakaldo/ES, 2Neuromuscular Disorders Group/Barakaldo/ES

PS1Group1-109 **PHENOTYPE-GENOTYPE RELATIONS IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 1**

*K. Mul*1, R.J. Lemmers2, C.G. Horlings1, P.J. Van Der Vliet2, M.A. Jonker3, N.C. Voermans1, S.M. Van Der Maarel4, B.G. Van Engelen1; 1Neurology/Nijmegen/NL, 2Human genetics/Leiden/NL, 3Health sciences/Nijmegen/NL, 4Human Genetics/Leiden/NL

PS1Group1-110 **SCN4A CAN ACT AS MODIFIER GENE IN PATIENTS WITH MYOTONIC DYSTROPHY TYPE 2**

A. Binda1, L.V. Renna2, F. Bose'3, E. Brigonzi4, A. Botta5, R. Valaperta6, B. Fossati7, I. Rivolta1, G. Meola4, *R. Cardani*2; 1School of Medicine and Surgery/Monza/IT, 2Laboratory of Muscle Histopathology and Molecular Biology/San Donato Milanese (mi)/IT, 3Laboratory of Muscle Histopathology and molecular biology/San Donato Milanese (mi)/IT, 4Department of Biomedical Sciences for Health/San Donato Milanese (mi)/IT, 5Department of Biomedicine and Prevention/Rome/IT, 6Research Laboratories/San Donato Milanese (mi)/IT, 7Department of Neurology/San Donato Milanese (mi)/IT

PS1Group1-111 **AAV-SERCA2A EXPRESSION AMELIORATED CARDIOMYOPATHY IN THE MDX MOUSE MODEL OF DUCHENNE MUSCULAR DYSTROPHY**

*Y. Yue*1, N. Wasala1, W. Lostal1, L. Wasala1, N. Niranjan2, R. Hajjar3, G. Babu2, D. Duan1; 1Molecular Microbiology and Immunology (Medical Science Bldg)/Columbia, Mo/US, 2Cell Biology and Molecular Medicine/Newark/US, 3/New York/US

PS1Group1-112 **QUANTITATIVE ANALYSIS OF THIGH MUSCLE BUNDLES OF PATIENTS WITH MYOTONIC DYSTROPHY TYPE 1 (DM1), USING CT IMPAIRMENT RATIO**

*T. Nakayama*1, S. Kuru2; 1Department of Neurology, Division of Neuromuscular Diseases/Yokohama/JP, 2Neurology/Suzuka/JP

PS1Group1-113 **LIMB-GIRDLE MUSCULAR DYSTROPHY IN THE CZECH REPUBLIC**

*K. Stehlikova*1, J. Zidkova1, K. Reblova2, M. Ruzicka2 R. Mazanec3, S. Vohanka4, J. Haberlova5, L. Fajkusova6; 1Centre of Molecular Biology and Gene Therapy/Brno/CZ, 2CEITEC/Brno/CZ, 3Neurology/Prague/CZ, 4Neurology/Brno/CZ, 56 Department of Child Neurology/Prague/CZ, 6/Brno/CZ

PS1Group1-114 **PULMONARY FUNCTION IN ADVANCED DUCHENNE MUSCULAR DYSTROPHY: ETEPLIRSEN-TREATED PATIENTS VERSUS A NATURAL HISTORY COHORT**

H. Gordish-Dressman1, E. Henricson2, L. Han3, A. Dugar3, C. McDonald2, Cooperative International Neuromuscular Research Group (cinrg)4, J. Dworzak; 1/Washington, DC/US, 2/Davis, CA/US, 3/Cambridge, MA/US, 4/US

PS1Group1-115 **CLINICAL OUTCOME STUDY OF DYSFERLINOPATHY: TEENAGE EXERCISE AS A POTENTIAL MODIFIER OF DISEASE SEVERITY**

*C. Semplicini*1, U. Moore2, M. Jacobs3, 4, R. Fernández-Torrón2, 5, J. Jang3, 4, M. James2, A. Mayhew2, L. Rufibach6, P. Mittal6, M. Eagle2, A. Cnaan4, P.G. Carlier7, A. Blamire8, H. Hilsden2, H. Lochmuller2, U. Grieben9, S. Spuler10, C. Tesi Rocha11, J.W. Day11, K.J. Jones12, D.X. Bharucha-Goebel13, E. Salort-Campana14, M. Harms15, A. Pestronk16, S. Krause17, O. Schreiber-Katz17, M.C. Walter17, C. Paradas18, J.-Y. Hogrel19, T. Stojkovic20, S. Takeda21, M. Mori-Yoshimura22, E. Bravver23, S. Sparks23, J.D. Manera24, 25, L. Bello26, E. Pegoraro1, J.R. Mendell27, K. Bushby2, V. Straub28; 1/Padova/IT, 2The John Walton Muscular Dystrophy Research Centre, Newcastle Hospitals Trust/Newcastle Upon Tyne/GB, 3Division of Biostatistic and Study Methodology, Children's National Health System/Dc, WA/US, 4Pediatrics, Epidemiology and Biostatistics/Washington Dc/US, 5Neuromuscular Area, Biodonostia Health Research Institute, Neurology Service, Donostia University Hospital/Donostia-San Sebastian/ES, 6/Seattle/US, 7AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 8Magnetic Resonance Centre, Institute for Cellular Medicine/Newcastle Upon Tyne/GB, 9Charite Muscle Research Unit, a joint cooperation of the Charité Medical Faculty and the Max Delbrück Center for Molecular Medicine/Berlin/DE, 10Charite Muscle Research Unit/Berlin/DE, 11Department of Neurology and Neurological Sciences/Stanford, CA/US, 12Institute for Neuroscience and Muscle Research, Children's Hospital at Westmead/Sydney/AU, 13Department of Neurology/Washington Dc/US, 14Neuromuscular and ALS Center, La Timone Hospital/Marseille/FR, 15Department of Neurology/St. Louis, MO/US, 16Department of Neurology/St. Louis, Mo/US, 17Friedrich-Baur Institute, Departement of Neurology/Munich/DE, 18Neuromuscular Unit, Department of Neurology/Sevilla/ES, 19Institut de Myologie/Paris/FR, 20AP-HP, G.H. Pitié-Salpêtrière 47-83, Boulevard de l'Hôpital/Paris/FR, 21/Tokyo/JP, 22Department of Neurology/Tokyo/JP, 23/Charlotte, Nc/US, 24Neuromuscular Unit, Neurology Department/Barcelona/ES, 25/Barcelona/ES, 26Department of Neuroscience/Padova/IT, 27/Columbus/US, 28John Walton Muscular Dystrophy Research Centre/Newcastle Upon Tyne/GB

PS1Group1-116 **GLYCOGENOSIS TYPE V (MCARDLE DISEASE): THERAPY WITH VITAMIN B6**

A. Saak, H. Reichmann, J. Schaefer; Neurology/Dresden/DE

PS1Group1-117 **MASSIVE INCREASE IN CARDIAC TROPONIN T WITHOUT CARDIAC INVOLVEMENT IN NECROTIZING MYOPATHY WITH ANTI-HMGCR-ANTIBODIES**

A. Saak, J. Schaefer, S. Jackson; Neurology/Dresden/DE

PS1Group1-118 **RESULTS OF NORTH STAR AMBULATORY ASSESSMENTS IN THE ACT DMD TRIAL IN IN PATIENTS WITH NMDMD**

*F. Muntoni*1, P. Trifillis2, G.L. Elfring2, S.W. Peltz2, C. McDonald3; 1/London/GB, 2/South Plainfield, NJ/US, 3Physical Medicine and Rehabilitation/Sacramento, CA/US

PS1Group1-119 **NONSENSE AND SINGLE NUCLEOTIDE FRAMESHIFT MUTATIONS IN BECKER MUSCULAR DYSTROPHY**

M. Traverso1, C. Panicucci1, M. Catteruccia2, P. Broda1, F. Madia3, G. Pozzolini3, M. Derchi4, A. Torella5, C. Bruno6, F. Zara3, V. Nigro5, C. Minetti1, A. D'Amico2, *C. Fiorillo*1, 7; 1Pediatric Neurology and Muscular

Disorders/Genoa/IT, 2Unit of Neuromuscular Disorders, Laboratory of Molecular Medicine/Rome/IT, 3Neurogenetic Laboratory/Genoa/IT, 4Cardiology/Genoa/IT, 5Precision Medicine/Naples/IT, 6Centre of Myology/Genoa/IT, 7Neuroscience/Genoa/IT

PS1Group1-120 **DEVELOPMENT OF A PROGNOSTIC MODEL FOR 1-YEAR CHANGE IN TIMED 4 STAIR-CLIMB IN DUCHENNE PATIENTS**

*N. Goemans*1, M. Vanden Hauwe1, J. Signorovitch2, 3, G. Sajeev2, Z. Yao2, M. Jenkins2, E. McDonald2, I. Dieye2, S.J. Ward3; 1Neuromuscular Reference Centre, Department of Paediatrics and Child Neurology/Leuven/BE, 2/Boston/US, 3/Cambridge/US

PS1Group1-121 **AMBULATORY ELECTROCARDIOGRAPHIC LONGITUDINAL STUDY IN THE GRMD DOG MODEL OF DUCHENNE MUSCULAR DYSTROPHY**

I. Barthélémy, X. Cauchois, S. Blot; U955 - IMRB, Inserm, Ecole Nationale Vétérinaire d'Alfort, UPEC/Maisons-Alfort/FR

PS1Group1-122 **MIMICS OF INCLUSION BODY MYOSITIS: CASE PRESENTATIONS AND IDENTIFICATION OF TYPICAL PITFALLS**

R. Zeng, S. Glaubitz, K. Schmidt, P.-O. Carstens, J. Schmidt, J. Zschüntzsch; Department of Neurology/Göttingen/DE

PS1Group1-123 **LONG-TERM PULMONARY FUNCTION IN NON-AMBULATORY PATIENTS WITH NMDMD TREATED WITH ATALUREN**

C. McDonald1, *E. Mercuri*2, F. Muntoni3, K. Selby4, F. Jin5, G. Panaghie-Meltzer5, P. Trifillis5, M. Souza5, S.W. Peltz5, M. Tulinius6; 1Physical Medicine and Rehabilitation/Sancramento, CA/US, 2/Rome/IT, 3/London/GB, 4/Vancouver, BC/CA, 5/South Plainfield, NJ/US, 6/Gothenburg/SE

PS1Group1-124 **USE OF A >/= 5-SECOND THRESHOLD IN BASELINE TIME TO STAND FROM SUPINE TO PREDICT PROGRESSION IN DMD**

C. McDonald1, M. Souza2, G.L. Elfring2, P. Trifillis2, J. McIntosh2, S.W. Peltz2, *E. Mercuri*3; 1Physical Medicine and Rehabilitation/Sacramento, CA/US, 2/South Plainfield, NJ/US, 3/Rome/IT

PS1Group1-125 **NOVEL THERAPEUTIC PERSPECTIVES FOR SARCOGLYCANOPATHY, IN VITRO AND IN VIVO STATE OF THE ART**

*D. Sandona*1, R. Sacchetto1, E. Bianchini1, M. Carotti1, M. Soardi1, C. Fecchio2; 1/Padova/IT, 2Biomedical science/Padova/IT

PS1Group1-126 **ATALUREN IN PATIENTS AGED ≥ 2 to**

C. Tian1, R. Kong2, F. Jin2, E. O'Mara2, P. Trifillis2, *J. McIntosh*2, J.B. Renfroe3; 1Neurology/Cincinatti, OH/US, 2/South Plainfield, NJ/US, 3/Gulf Breeze, FL/US

PS1Group1-127 **A FEMALE CARRIER OF BECKER MUSCULAR DYSTROPHY PRESENTING WITH A MYOPATHY WITH PIPESTEM CAPILLARIES**

*G. Cosentino*1, F. Brighina1, B. Fierro1, L. Pilati1, M. Mirabella2, C. Rodolico3; 1/Palermo/IT, 2/Roma/IT, 3/Messina/IT

PS1Group1-128 **URINARY TITIN IS A NON-INVASIVE BIOMARKER TO DIAGNOSE DUCHENNE MUSCULAR DYSTROPHY EVEN IN ADVANCED STAGE**
*H. Awano*1, Y. Ishikawa2, Y. Ishikawa2, M. Matsumoto1, M. Nagai1, T. Shirakawa3, N. Maruyama4, Y.-I. Nabeshima5, K. Iijima1, M. Matsuo3; 1Pediatrics/Kobe/JP, 2/Yakumo/JP, 3Physical Therapy/Kobe/JP, 4/Fujioka/JP, 5Laboratory of Molecular Life Science/Kobe/JP

PS1Group1-129 **A NOVEL MUSCLE PHENOTYPE IN A PATIENT WITH TROPOMYOSIN-RECEPTOR KINASE-FUSED GENE (TFG) DISEASE**
*N.N. Madigan*1, J.A. Tracy1, W.J. Litchy1, Z. Niu2, M. Milone1; 1Neurology/Rochester, MN/US, 2Laboratory Genetics & Genomics/Rochester, MN/US

PS1Group1-130 **GLYCOGEN STORAGE DISEASE TYPE IV PRESENTING AS CONGENITAL MYOPATHY WITH CONTRACTURES AND RIGID SPINE**
*M.C. Walter*1, S. Wenninger1, A. Abicht2; 1Dept. of Neurology, Ludwig-Maximilians-University/Munich/DE, 2/Munich/DE

PS1Group1-131 **STATIN-ASSOCIATED AUTOIMMUNE NECROTIZING MYOPATHY**
*S.H. Ahn*1, J.-J. Sung1, Y.-H. Hong2, K.H. Kwun1, J.A. Kim1, J.-Y. Shin2, S.M. Kim1, A.W. Kim1; 1Neurology/Seoul/KR, 2/Seoul/KR

PS1Group1-132 **PHENOTYPIC HETEROGENEITY IN THREE PATIENTS WITH M.3243A>G MUTATION**
*B. Burnyte*1, K. Grigalioniene1, A. Vaitkevicius2, D. Petroska3, 4, L. Cimbalistiene1, V. Kucinskas1, A. Utkus1; 1Institute of Biomedical Sciences of the Faculty of Medicine/Vilnius/LT, 2Institute of Clinical Medicine of the Faculty of Medicine/Vilnius/LT, 3Department of Pathology/Vilnius/LT, 4Affiliate of Vilnius University Hospital Santaros Klinikos/Vilnius/LT

PS1Group1-133 **DOUBLE TROUBLE: A CHILD WITH DUCHENNE MUSCULAR DYSTROPHY AND NOONAN SYNDROME**
*Y.J. Lee*1, H. Jang1, J.Y. Park2, J.S. Park3; 1Pediatrics/Daegu/KR, 2Pathology/Daegu/KR, 3Neurology/Daegu/KR

PS1Group1-134 **THE HETEROZYGOUS R155C VCP MUTATION: TOXIC IN HUMANS, HARMLESS IN MICE**
*C. Clemen*1, L. Winter2, K.-H. Strucksberg3, C. Berwanger1, M. Türk4, L. Eichinger3, The German Mouse Clinic Consortium5, R. Schröder6; 1Department of Neurology, Heimer Institute for Muscle Research/Bochum/DE, 2Center for Anatomy and Cell Biology/Vienna/AT, 3Institute of Biochemistry, Medical Faculty/Cologne/DE, 4Department of Neurology/Erlangen/DE, 5/Munich/DE, 6Neuropathology/Erlangen/DE

PS1Group1-135 **ADAPTIVE PROTEIN QUALITY CONTROL RESPONSE IN DESMINOPATHY SKELETAL MUSCLE CELLS AND TISSUE**
*C. Clemen*1, L. Winter2, C. Berwanger1, M. Spörrer3, U. Schlötzer-Schrehardt4, R. Schröder5; 1Department of Neurology, Heimer Institute for Muscle Research/Bochum/DE, 2Center for Anatomy and Cell Biology/Vienna/AT, 3Department of Physics/Erlangen/DE, 4Department of Opthalmology/Erlangen/DE, 5Neuropathology/Erlangen/DE

PS1Group1-136 **CLINICAL BACKGROUND OF 94 ADULT PATIENTS WHO RECOGNIZED NEMALIN RODS IN MUSCLE TISSUE**
*A. Hashiguchi*1, K. Kodama2, I. Higuchi3, H. Takashima2; 1Neurology, Neurological Disease Center/Kagoshima City/JP, 2Department of Neurology and Geriatrrics/Kagoshima/JP, 3Physical Therapy foundation course/Kagoshima/JP

PS1Group1-137 **P62 IMMUNOSTAINING COULD HELP IN DIFFERENTIATING POLYMYOSITIS FROM SIBM**
*J.C. Milisneda*1, C. Jou2, I. Pinal-Feranndez3, J.M. Grau4; 1Internal Medicine/Barcelona/ES, 2/Barcelona/ES, 3National Institute of Arthritis and Musculoskeletal and Skin Diseases/Bethesda, Maryland, AL/US, 4Internal Medicine Service/Barcelona/ES

PS1Group1-138 **HOMOZYGOSITY OF THE AUTOSOMAL DOMINANT VCP P.ARG159HIS MUTATION**
*W. De Ridder*1, T. Deconinck1, P. De Jonghe1, K. Johnson2, A. Töpf2, M. Bertoli2, L. Phillips2, D. Macarthur3, J. Baets1; 1Neurogenetics Group, VIB-UAntwerp Center for Molecular Neurology/Antwerp/BE, 2John Walton Muscular Dystrophy Research Center/Newcastle Upon Tyne/GB, 3/Cambridge, MA/US

PS1Group1-139 **GENOTYPE CHARACTERIZATION OF CZECH PATIENTS WITH FACIOSCAPULOHUMERAL DYSTROPHY**
*J. Zidkova*1, S. Vohanka2, R. Mazanec3, L. Fajkusova4; 1Centre of Molecular Biology and Gene Therapy/Brno/CZ, 2Neurology/Brno/CZ, 3Neurology/Prague/CZ, 4/Brno/CZ

PS1Group1-140 **OCULOPHARYNGEAL MUSCLE WEAKNESS AFTER TREATMENT WITH CHECKPOINT INHIBITORS: PATHOLOGY IS BEYOND THE NEUROMUSCULAR JUNCTION**
*A.C. Mehrabyan*1, A. Ahmed2, C. Specht3; 1Neurology/Chapel Hill, /US, 2Neurology/Hershey, PA/US, 3/Hershey, PA/US

PS1Group1-141 **A CASE OF CENTRAL CORE DISEASE WITH NOVEL RYR1 MUTATION IN KOREAN PATIENT**
*J.-Y. Shin*1, C. Huh2, S.H. Ahn3, K.H. Kwun1, J.A. Kim1, A.W. Kim1, Y.-H. Hong2, J.-J. Sung1; 1Neurology/Seoul/KR, 2/Seoul/KR, 3Neurology /Seoul/KR

PS1Group1-142 **MYOADENILATE DEAMINASE DEFICIENCY IN PATIENTS WITH MYALGIA**
G. Siciliano, C. Simoncini, G. Ricci; Department of Clinical and Experimental Medicine/Pisa/IT

PS1Group1-143 **A PHASE IIA STUDY OF TAS-205, A NOVEL INHIBITOR OF HEMATOPOIETIC PROSTAGLANDIN D SYNTHASE, IN DUCHENNE MUSCULAR DYSTROPHY**
*T. Matsumura*1, H. Komaki2, S. Kuru3, T. Nakayama4, S. Takeda5; 1Neurology/Toyonaka/JP, 2Child Neurology/Kodaira/JP, 3Neurology/Suzuka/JP, 4Neurology/Yokohama/JP, 5/Kodaira/JP

PS1Group1-144 **USE OF HUMAN INDUCED PLURIPOTENT STEM CELLS FOR MODELLING SKELETAL MUSCULAR DEFECTS ASSOCIATED TO MYOTONIC DYSTROPHY TYPE 1**

*J. Tahraoui*1, L. Lesueur2, C. Pinset2, X. Nissan2, D. Furling3, C. Martinat2; 1I-Stem/Corbeil Essonnes/FR, 2I-STEM/Corbeil Essonnes/FR, 3Institut de Myologie/Paris/FR

PS1Group1-145 **A CASE OF FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 2 WITH NOVEL FRAMESHIFT MUTATION OF SMCHD1 GENE IN KOREA**

*J.H. Lee*1, M.-W. Seong2, C.-S. Ki3, Y.C. Choi1; 1Department of Neurology/Seoul/KR, 2Department of Laboratory Medicine/Seoul/KR, 3Department of Laboratory Medicine and Genetics/Seoul/KR

PS1Group1-146 **ASYMMETRIC WEAKNESS IN HMGCOA REDUCTASE ANTIBODY NECROTIZING MYOPATHY**

M. Soni; Neurological Sciences/Chicago, IL/US

PS1Group1-147 **CAN NECK FLEXION WEAKNESS PREDICT CHANGES IN SWALLOWING AND COUGH PEAK FLOW IN PATIENTS WITH MYOTONIC DYSTROPHY TYPE 1?**

*C. Massey*1, J.E. Allen2, U. Vivekananda3, N. Nikolenko4, C. Jimenez-Moreno4, C. Turner5; 1Therapy and Rehabilitation/Bg/GB, 2Therapy and Rehabilitation /3bg/GB, 3/Bu/GB, 4/Bz/GB, 5Institute of Neurology/Bg/GB

PS1Group1-148 **THE ADDITIONAL VALUE OF HISTOPATHOLOGICAL FASCIA EXAMINATION IN DIAGNOSING MYOSITIS**

A. Van Der Kooi; Neurology/Amsterdam/NL

Sunday, July 8, 2018

17:15 - 18:30 **Guided Poster Session**
Room: Mezzanine Floor Gallery and Foyers

- Topic Group 3: Peripheral Neuropathy, Cranial Nerves, Mononeuropathies: Clinical Features, Pathophysiology, Therapy
- Topic Group 5: Novel Diagnostic Methods in Neuromuscular Diseases and Basic Sciences in Neuromuscular Diseases
- Topic Group 6: Cancer related Disorders and General Diseases
- Topic Group 9: Miscellaneous

PS2Group3-001 **A TALE OF TWO CHARCOT'S JOINTS: LONDON AND DAR-ES-SALAAM**

C.Y. Kok, H. Manji; MRC Neuromuscular Centre/London/GB

PS2Group3-002 **HETEROGENEITY OF THE PATIENT PATHWAY AND CHARACTERISTICS FOR HATTR AMYLOIDOSIS: PERSPECTIVES FROM CENTRAL AND EASTERN EUROPE**
*Y. Parman*1, I. Tournev2, D. Coriu3, M. Arad4, M. Lipowska5, S. Nikitin6, S. Sarafov2, J. Zidar7; 1/Istanbul/TR, 2/Sofia/BG, 3/Bucharest/RO, 4/Tel-Hashomer/IL, 5/Warsaw/PL, 6/Moscow/RU, 7/Ljubljana/SI

PS2Group3-003 **AUTONOMIC AND SENSORY NEUROPATHY: CHALLENGES IN THE ETIOLOGY AND TREATMENT OF A PEDIATRIC CASE**
M. Sampaio1, C. Garrido1, M. Santos1, *A. Sousa*2, S. Figueiroa1; 1Neuropediatrics/Porto/PT, 2Neurophysiology/-/PT

PS2Group3-004 **EXTENSIVE GENETIC ANALYSIS IN A TAIWANESE COHORT WITH CHARCOT-MARIE-TOOTH DISEASE**
K.-P. Lin1, Y.-C. Liao2, *Y.-C. Lee*2; 1/Taipei/TW, 2Neurology/Taipei/TW

PS2Group3-005 **IMPACT OF PATISIRAN ON AUTONOMIC NEUROPATHY IN HEREDITARY TRANSTHYRETIN-MEDIATED AMYLOIDOSIS PATIENTS**
*M. Mauermann*1, D. Adams2, A. Gonzalez-Duarte3, T. Coelho4, C.-C. Yang5, M. Polydefkis6, A. Kristen7, I. Tournev8, H. Schmidt9, J. Berk10, K.-P. Lin11, P. Gandhi12, M. Sweetser12, M. White12, J. Gollob12, O. Suhr13; 1/Rochester/US, 2Neurology Department/Le Kremlin Bicetre/FR, 3/Salvador Zubiran/MX, 4/Porto/PT, 5/Taipei/TW, 6/Baltimore, MD/US, 7/Heidelberg/DE, 8/Sofia/BG, 9/Muenster/DE, 10/Boston, MA/US, 11Neurology/Taipei/TW, 12/Cambridge, MA/US, 13Department of Public Health and Clinical Medicine/Umea/SE

PS2Group3-006 **THE FUNCTIONAL AND STRUCTURAL EVALUATION OF SMALL FIBERS IN ASYMPTOMATIC PATIENTS WITH VAL30MET MUTATION**
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PS2Group3-007 **CLINICAL AND ELECTROPHYSIOLOGICAL FEATURES OF AMAN FROM THE SONORAN OUTBREAK**

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PS2Group3-008 **METABOLIC SYNDROME COMPONENTS AND NEUROLOGIC OUTCOMES IN A BARIATRIC SURGERY POPULATION**
B.C. Callaghan, E. Villegas-Umana, E. Chant, E. Reynolds, M. Banerjee, E. Feldman; /Ann Arbor/US

PS2Group3-009 **A SUCCESSFUL TREATMENT OF IDIOPATHIC BRACHIAL NEURITIS WITH SONO-GUIDED INJECTION AND LOW DOSE STEROID THERAPY**
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PS2Group3-010 **APOLLO PHASE 3 STUDY: IMPACT OF BASELINE NEUROPATHY SEVERITY ON RESPONSE TO PATISIRAN**
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PS2Group3-011 **CLINICAL SPECTRUM OF HEREDITARY SPASTIC PARAPARESIS BY MUTATION IN KIF1A GENE**
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PS2Group3-013 **THE EFFECT OF REHABILITATION ON SLEEP, RESPIRATORY AND LIFE QUALITY**
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PS2Group3-014 **MULTIFOCAL MOTOR NEUROPATHY WITH CONDUCTION BLOCK: A CASE SERIES**
A.A. Alshareef; Neurology/Jeddah/SA

PS2Group3-015 **PHARMACOKINETICS AND SAFETY OF A SELECTIVE ANTIBODY-SCAVENGING GLYCOPOLYMER FOR THE TREATMENT OF ANTI-MAG NEUROPATHY**
*D. Demeestere*1, B. Aliu1, W. Heusermann1, B. Ernst1, P. Hänggi2, R. Herrendorff2; 1Institute of Molecular Pharmacy/Basel/CH, 2R&D/Basel/CH

PS2Group3-016 **A NOVEL MISSENSE MUTATION OF TRANSTHYRETIN CAUSING AMYLOIDOSIS**
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PS2Group3-017 **CHRONIC MOTOR AXONAL NEUROPATHIES: A CHALLENGING DIAGNOSIS**
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PS2Group3-018 **QUALITY OF LIFE IN A CLINICAL STUDY OF MAINTENANCE TREATMENT OF CIDP WITH IGPRO20: THE PATH STUDY**

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PS2Group3-019 **FEASIBILITY OF SWITCHING FROM INTRAVENOUS TO SUBCUTANEOUS IG THERAPY IN CIDP: PATH TRIAL RESULTS VERSUS CLINICAL EXPERIENCE**

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PS2Group3-020 **TWO NOVEL VARIANTS IN THE SLC25A46 GENE CAUSING OPTICAL ATROPHY AND PERIPHERAL NEUROPATHY IN CZECH SIBLINGS**

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PS2Group3-021 **GENOME-WIDE DNA METHYLATION PROFILING OF HUMAN DIABETIC PERIPHERAL NEUROPATHY**

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PS2Group3-022 **TRANSCRIPTIONAL SIGNATURE OF DIABETIC PERIPHERAL NEUROPATHY CONSERVED ACROSS HUMAN AND MOUSE**

E. Feldman; /Ann Arbor/US

PS2Group3-023 **RESTABILISATION TREATMENT AFTER IVIG WITHDRAWAL IN CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY: THE PATH STUDY RESULTS**

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PS2Group3-024 **EFFICACY AND SAFETY OF INTRAVENOUS IMMUNOGLOBULIN IGPRO10 IN CIDP: COMBINED ANALYSIS OF THE PRIMA AND PATH STUDIES**

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PS2Group3-025 **A FATAL CASE OF HEPATITIS E-ASSOCIATED GUILLAIN-BARRE SYNDROME**

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PS2Group3-026 **PUTATIVE DIGENIC INHERITANCE OF CHARCOT-MARIE-TOOTH DISEASE INVOLVING TWO NOVEL HETEROZYGOUS MUTATIONS IN MARS AND HARS**

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PS2Group3-027 **A RANDOMIZED, BLINDED, LIFESTYLE INTERVENTION STUDY IMPROVES THE EXPIRATION:INSPIRATION RATIO IN DIABETIC NEUROPATHY**

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PS2Group3-028 **PREDICTORS OF EARLY RETIREMENT IN PATIENTS WITH CHRONIC INFLAMMATORY DEMYELINATING POLYRADICULONEUROPATHY**

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PS2Group3-029 **NEUROPATHY AS AN ADVERSE EFFECT OF VASCULAR ENDOTHELIAL GROWTH FACTOR TYROSINE KINASE INHIBITORS, A META-ANALYSIS**

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PS2Group3-030 **AXONAL FUNCTION PREDICTS RESPONSE TO SUBCUTANEOUS IMMUNOGLOBULIN IN CIDP: THE PATH STUDY**

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PS2Group3-031 **ELECTROPHYSIOLOGICAL TESTING IN PATIENTS WITH CIDP TREATED WITH SUBCUTANEOUS IMMUNOGLOBULIN: THE PATH STUDY**
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PS2Group3-032 **CHRONIC RELAPSING INFLAMMATORY OPTIC NEUROPATHY (CRION): A MANIFESTATION OF MYELIN OLIGODENDROCYTE GLYCOPROTEIN ANTIBODIES**
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PS2Group3-033 **PREVALENCE AND RISK FACTORS OF CARPAL TUNNEL SYNDROME IN OYSTER-SHUCKERS**
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PS2Group3-034 **NEXT GENERATION SEQUENCING TECHNOLOGIES IN THE GENETIC DIAGNOSIS OF EARLY ONSET HEREDITARY SPASTIC PARAPLEGIAS**
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PS2Group3-035 **A CASE OF SEROPOSITIVE NEUROMYELITIS OPTICA IN A PATIENT WITH CO-EXISTING MYASTHENIA GRAVIS AND SYSTEMIC LUPUS ERYTHEMATOSUS**
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PS2Group3-036 **NERVE ULTRASOUND ASSESSMENT IN A NOVEL MUTATION C.379DELG (PALA127LEUFS*52) IN DRP2 GENE**
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PS2Group3-037 **PHRENIC NERVE DEMYELINATION CAUSING ACUTE RESPIRATORY FAILURE IN MADSAM**
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PS2Group3-038 **BACLOFEN, NALTREXONE AND SORBITOL ALL CONTRIBUTE TO THE EFFICACY OF PXT3003 IN CMT1A RATS**
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PS2Group3-039 **IDEAL TIME FOR A REPEAT CONDUCTION TO SUBTYPE VARIANTS IN GUILLAIN BARRE SYNDROME**
A.K. Meena, R. Chepuru, S. Sarva, S. Yareeda, N. Mathukomali; Neurology/Hyderabad/IN

PS2Group3-040 **COST OF ILLNESS IN CHARCOT-MARIE-TOOTH NEUROPATHY: RESULTS FROM GERMANY**
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PS2Group3-041 **CLINICAL HETEROGENEITY OF ANTI-GM2-GANGLIOSIDE ANTIBODY SYNDROME**
J.S. Bae, *S.M. Yoon*, S.K. Shim; Neurology/Seoul/KR

PS2Group3-042 **LATE-ONSET AXONAL CHARCOT-MARIE-TOOTH ASSOCIATED WITH AUTOSOMAL RECESSIVE MME MUTATIONS**
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PS2Group3-043 **POPULATION PHARMACOKINETICS (PK) OF INVESTIGATIONAL PATISIRAN IN HEALTHY VOLUNTEERS AND IN PATIENTS**
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PS2Group3-044 **CHARACTERIZATION OF NEURONAL MOLECULAR MECHANISMS UNDERLYING CMT2Z NEUROPATHY**
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PS2Group3-045 **INCREASED DEACETYLATION OF PROTEINS BY SIRTUIN 1 PROTEIN OVER EXPRESSION REVERSES T2D PERIPHERAL NEUROPATHY**
J. Russell, K. Chandrasekaran, C.-Y. Ho, P. Kumar, S.S. Reddy; /Baltimore/US

PS2Group3-046 **PGC-1α REGULATES NEURONAL RESPITRATORY CHAIN SUPERCOMPLEXES IN MITOCHONDRIA**
J. Russell, J. Choi, M. Salimian, S.S. Reddy; /Baltimore/US

PS2Group3-047 **THE VALIDITY OF SUDOSCAN IN SCREENING OF UPPER EXTREMITY NEUROPATHIES: A PILOT STUDY**
G.-Y. Park, S. Im, H.J. Koo, Y. Jang, C.S. Chae; Rehabilitation Medicine/Bucheon-si/KR

PS2Group3-048 **IPSC MOTOR NEURONS FROM AN X-LINKED CMT TYPE 6 PATIENT AS A MODEL FOR STUDYING AXONAL DEGENERATION AND DEVELOPING THERAPIES**
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PS2Group3-049 **PARANODAL ANTIBODIES IN AUSTRIAN PATIENTS WITH ACUTE ONSET INFLAMMATORY NEUROPATHY**
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PS2Group3-050 **ULNAR NERVE CONDUCTION STUDY USING THE FIRST DORSAL INTEROSSEOUS MUSCLE RECORDING IN HEALTHY KOREAN SUBJECTS**
J.-Y. Cho, H.J. Lee; Neurology/Goyang-si, Gyeonggi-do/KR

PS2Group3-051 **PATIENT ASSISTED INTERVENTION FOR NEUROPATHY: COMPARISON OF TREATMENT IN REAL LIFE SITUATIONS (PAIN-CONTROLS)**
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PS2Group3-052 **A NEW MUTATION IN MORC 2 CAUSES SCAPULOPERONEAL CHARCOT-MARIE-TOOTH NEUROPATHY: DAVIDENKOW SYNDROME**
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PS2Group3-053 **AUTOPHAGIC NEUROMYOPATHY CAUSED BY P.R140G HEAT SHOCK PROTEIN B1 (HSPB1) MUTATION**
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PS2Group3-054 **HEREDITARY TRANSTHYRETIN AMYLOIDOSIS MIMICKING REFRACTORY CHRONIC INFLAMMATORY DEMYELINATING POLYRADICULONEUROPATHY**
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PS2Group3-055 **TRANSTHYRETIN REDUCTION WITH PATISIRAN IN THE APOLLO PHASE 3 STUDY**
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8/Sofia/BG, 9/Muenster/DE, 10/Boston, MA/US, 11/Cambridge, MA/US, 12Department of Public Health and Clinical Medicine/Umea/SE

PS2Group3-056 **INFLAMMATORY NEUROPATHY AFTER IMMUNE CHECKPOINT THERAPY**
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PS2Group3-057 **BENEFIT-RISK PROFILE OF INTRAVENOUS IMMUNOGLOBULIN (IVIG) AND SUBCUTANEOUS IMMUNOGLOBULIN (SCIG) IN CIDP: THE PATH STUDY**
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PS2Group3-058 **EPIDEMIOLOGY OF HEREDITARY MOTOR-SENSORY NEUROPATHIES IN THE POPULATION OF THE REPUBLIC OF BASHKORTOSTAN**
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PS2Group3-059 **INFLUENCE OF THE EXTRACT OF SUCUPIRA BRANCA (PTERODON EMARGINATUS VOGEL) IN SURAL NERVE OF DIABETIC RATS**
B.M. Bertassoli1, A.S. Faiola1, J. Armando Jr1, G. Petri1, J.F.R. Santos1, F.L.A. Fonseca2, *A.A.S. Carvalho*3, D. Feder1; 1Pharmacology/Santo Andre/BR, 2Laboratory Analysis/Santo Andre/BR, 3Neurology/Santo Andre/BR

PS2Group3-060 **EXPANDING THE PHENOTYPIC SPECTRUM OF GARS MUTATIONS**
G. Ricci, V. Boczonadi, B. Bansagi, R. Horvath; Institute of Genetic Medicine/Newcastle/GB

PS2Group3-061 **REDDISH SKIN COLOR CHANGE IN A PATIENT WITH COMPRESSIVE RADIAL NEUROPATHY**
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PS2Group3-062 **UNCLASSIFIED CONGENITAL AXONAL NEUROPATHY IN GIPSY FAMILLIES IN PORTUGAL**
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PS2Group3-063 **ETIOLOGY AND OUTCOME IN NEUROMUSCULAR PATIENTS PRIMARILY PRESENTING WITH DIAPHRAGMATIC DYSFUNCTION**
*M. Türk*1, I. Weber2, G. Vogt-Ladner2, R. Schröder3, M. Winterholler2; 1Department of Neurology/Erlangen/DE, 2Department of Neurology/Schwarzenbruck/DE, 3Neuropathology/Erlangen/DE

PS2Group3-064 **IQYMUNE® IS EFFECTIVE AS MAINTENANCE TREATMENT FOR MMN: A RANDOMISED, DOUBLE-BLIND, CROSS-OVER STUDY VERSUS KIOVIG®**

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PS2Group3-065 **ANTI-MAG TITERS PRE/POST DEGLYCOSYLATION IN PATIENTS WITH IGM NEUROPATHY: CORRELATION WITH CLINICAL PHENOTYPE IN 8 CASES**

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PS2Group3-066 **ULTRASONOGRAPHY IS USEFUL IN EVALUATION FOR ULNAR NEUROPATHIES WITH NO LOCALIZATION IN ELECTROPHYSIOLOGICAL STUDIES**

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PS2Group3-067 **CLINICAL AND GENETIC ANALYSIS OF HEREDITARY PERIPHERAL NEUROPATHY IN EGYPTIAN POPULATION**

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PS2Group3-068 **THE USEFULNESS OF NERVE CONDUCTION STUDIES IN CHEIRALGIA PARESTHETICA**

I.S. Joo; Neurology/Suwon/KR

PS2Group3-069 **BACLOFEN, NALTREXONE AND SORBITOL ALL CONTRIBUTE TO PXT3003-INDUCED MYELINATION IN CMT1A DRG CO-CULTURES**

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PS2Group3-070 **AN AUTOSOMAL DOMINANT FAMILY WITH UNCOMPLICATED SPASTIC PARAPLEGIA DUE TO A STOP MUTATION IN HARS**

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PS2Group3-071 **CONDUCTION STUDIES OF PHRENIC NERVE IN HEALTHY CHILDREN**

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PS2Group3-072 **FIBER TRACTOGRAPHY OF FACIAL NERVE: IMPLICATIONS FOR HEMIFACIAL SPASM**

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PS2Group3-073 **NEUROMYOPATHY CAUSED BY LONG TERM COLCHICINE THERAPY**

M.Y. Chun, J.H. Lee, K.-D. Park; Neurology/Seoul/KR

PS2Group3-074 **CHARCOT MARIE TOOTH DISEASE TYPE 2CC DUE TO A MUTATION IN THE NEUROFILAMENT-HEAVY GENE IN A GERMAN FAMILY**

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PS2Group3-075 **ISOLATED ABDUCENS PALSY AS TOLOSA-HUNT SYNDROME IN A SYSTEMIC LUPUS ERYTHEMATOSUS PATIENT**
V. Serban; /Cluj/RO

PS2Group3-076 **MULTIFOCAL MOTOR NEUROPATHY IN AUSTRIA: A NATIONWIDE SURVEY**
E.-M. Oberreiter1, S. Quasthoff2, M. Erdler3, R. Topakian4, S. Grinzinger5, F. Zimprich6, K. Stieglbauer7, J. Wanschitz1, S. Höger2, K. Berek8, C. Thaler-Wolf9, N. Embacher10, J. Jecel3, I. Heß-Eberle5, W. Kleindienst5, M. Huemer11, E. Laich12, D. Oel4, W. Oertl7, E. Lenzenweger7, *W. Löscher*1; 1/Innsbruck/AT, 2/Graz/AT, 3/Vienna/AT, 4Neurology/Wels/AT, 5/Salzburg/AT, 6Department of Neurology/Vienna/AT, 7/Linz/AT, 8/Kufstein/AT, 9/Hall iT/AT, 10/St. Pölten/AT, 11/Schwarzach Im Pongau/AT, 12/Steyr/AT

PS2Group3-077 **AUTOSOMAL RECESSIVE CHARCOT MARIE TOOTH DISEASE: CLINICAL, ELECTROPHYSIOLOGY AND GENETIC SPECTRUM IN A TUNISIAN SERIES**
I. Kacem1, E. Ellouz2, M. Ben Djebara1, E. Leguern3, A. Gargouri1, *R. Gouider*4; 1/Tunis/TN, 2Neurology/Ibn Khaldoun Street Gabes/TN, 3/Paris/FR, 4Neurology/Manouba/TN

PS2Group3-078 **COLCHICINE INDUCED NEUROMYOPATHY IN A PATIENT USING CONCOMITANT DIURETICS**
Y.-D. Kim; Neurology/Incheon/KR

PS2Group3-079 **A CASE OF MULTIFOCAL MOTOR NEUROPATHY: COMPLEMENTARY ROLE OF ULTRASOUND**
*J.I. Suk*1, H.J. Rha2; 1Neurology/Daegu/KR, 2/Daegu/KR

PS2Group3-080 **PHARMACOKINETICS OF PATISIRAN IN PATIENTS WITH HEREDITARY TRANSTHYRETIN-MEDIATED AMYLOIDOSIS**
X. Zhang, V. Goel, G. Robbie; /Cambridge, MA/US

PS2Group3-081 **ROLE OF THE ER STRESS TRANSCRIPTION FACTOR XBP1 IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1B**
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PS2Group3-082 **OBINUTUZUMAB, A NEW ANTI-CD20 ANTIBODY, IS ACTIVE AND EFFECTIVE IN ANTI-MAG ANTIBODY POLYNEUROPATHY**
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PS2Group5-001 **LATE-ONSET POMPE DISEASE ASSOCIATED WITH POLYNEUROPATHY**
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PS2Group5-002 **CORRELATION BETWEEN ELECTRICAL IMPEDANCE MYOGRAPHY AND TWO QUANTITATIVE ULTRASOUND PARAMETERS IN DUCHENNE MUSCULAR DYSTROPHY**

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PS2Group5-003 **PHYSIOLOGICAL DIFFERENCES IN SARCOLEMMAL EXCITABILITY MEASUREMENTS BETWEEN HUMAN MUSCLES**

R. Boland-Freitas, J. Lee, *K. Ng*; Neurology/St Leonards/AU

PS2Group5-004 **LABEL-FREE IMAGING OF ABNORMAL LIPID ACCUMULATION IN PERIPHERAL NERVES FROM FABRY DISEASE PATIENTS USING RAMAN SPECTROSCOPY**

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PS2Group5-005 **MODULATING EFFECTS OF UNSATURATED FATTY ACIDS ON GENE EXPRESSION OF MYOSIN HEAVY CHAIN CLASS I AND IIB IN C2C12 MYOCYTES**

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PS2Group5-006 **ELECTROPHYSIOLOGICAL DIAGNOSTIC OF NEUROMUSCULAR DISEASES IN NEW-BORNS, INFANTS AND TODDLERS**

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PS2Group5-007 **DISCOVERY OF PROTEIN BIOMARKERS FOR DYSFERLINOPATHY**

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Biochemistry and Microbiology/Victoria, Bc/CA, 25Department of Computer Science/Vancouver, Bc/CA

PS2Group5-008 **NERVE ULTRASOUND FOR THE IDENTIFICATION OF TREATMENT-RESPONSIVE CHRONIC NEUROPATHIES WITHOUT NERVE CONDUCTION ABNORMALITIES**
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PS2Group5-009 **MOTOR-UNIT NUMBER ESTIMATION IN THE ABDUCTOR POLICIS BREVIS MUSCLE OF PATIENTS WITH CARPAL TUNNEL SYNDROME**
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PS2Group5-010 **HIGH RESOLUTION NEUROSONOGRAPHY IN PATIENTS WITH CARPAL TUNNEL SYNDROME AND NORMAL CONTROLS**
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PS2Group5-011 **CAN WE EARLY DETECT CARDIAC DYSFUNCTION IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY?**
M. Marchel, J. Kochanowski, A. Serafin, B. Truszkowska, G. Opolski; Department of Cardiology/Warsaw/PL

PS2Group5-012 **PERIPHERAL NERVOUS SYSTEM DISORDERS IN MULTIPLE MYELOMA: RABAT CLINICAL NEUROPHYSIOLOGY DEPARTMENT EXPERIENCE**
L. Belarabi, N. Birouk, B. Kably, L. Errguig, H. Belaïdi, R. Ouazzani; Clinical Neurophysiology/Rabat/MA

PS2Group5-013 **CRAMPS, FASCICULATIONS AND MUSCLE FATIGABILITY DUE TO VITAMIN D DEFICIENCY**
L. Belarabi, N. Birouk, B. Kably, R. Ouazzani; Clinical Neurophysiology/Rabat/MA

PS2Group5-014 **NEXT-GENERATION SEQUENCING FOR MOLECULAR DIAGNOSIS IN NEUROMUSCULAR DISORDERS. RESULTS FROM 151 PATIENTS**
*D. Natera De Benito*1, J. Exposito Escudero2, D. Yubero2, L. González Quereda2, P. Gallano Petit2, A. Töpf3, C. Ortez4, L. Carrera-García2, A.L. Frongia2, C. Jou2, C. Jimenez Mallebrera2, A. Codina2, J. Colomer2, A. Nascimento2; 1Neuromuscular Disorders Unit/Barcelona/ES, 2/Barcelona/ES, 3John Walton muscular dystrophy research center/Newcastle/GB, 4Neuromuscular Unit/Barcelona/ES

PS2Group5-015 **TRANSCRANIAL MAGNETIC STIMULATION AS A DIAGNOSTIC AND PROGNOSTIC TOOL IN CHILDREN WITH SEQUELAE OF ACUTE TRANSVERSE MYELITIS**
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PS2Group5-016 **TARGETED METHYL-SEQ QUANTIFICATION BY NGS TECHNOLOGY FOR ROUTINE DIAGNOSTIC OF FSHD**
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PS2Group5-017 **FACIAL NERVE ULTRASOUND IN BELL'S PALSY**
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PS2Group5-018 **CGRP UPREGULATES MYOSIN HEAVY CHAIN TYPE I MESSENGER RNA THROUGH CALCINEURIN-IL-6-INDEPENDENT MANNER IN C2C12 CELLS**
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PS2Group5-019 **ANALYSIS OF EUROPEAN EARLY-ONSET MYASTHENIA GRAVIS GWAS SIGNALS IN AFRICAN GENOMES**
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PS2Group5-020 **RNASEQ IN URINE-DERIVED STEM CELLS IDENTIFIED THE EXPRESSION OF 264 NEUROMUSCULAR GENE TRANSCRIPTS**
M.S. Falzarano1, H. Osman1, R. Rossi1, R. Selvatici1, M. Neri1, F. Gualandi1, M. Fang2, Z. Lu2, A. Grilli3, S. Bicciato3, *A. Ferlini*1; 1Medical Sciences/Ferrara/IT, 2/Shenzhen/CN, 3Dipartimento di Scienze della Vita/Modena/IT

PS2Group5-021 **NEURONOPATHY IN SPINOCEREBELLAR ATAXIA TYPE 2: A NERVE ULTRASOUND STUDY**
*L. Pelosi*1, R. Iodice2, A. Antenora2, D. Kilfoyle1, E. Mulroy1, M. Rodrigues1, R. Roxburgh1, A. Filla2, F. Manganelli2, L. Santoro1; 1Neurology and Clinical Neurophysiology/Auckland/NZ, 2Neurosciences, Reproductive Sciences and Odontostomatology/Naples/IT

PS2Group5-022 **NERVE ULTRASOUND IN POEMS REVEALS PROXIMAL AND DISTAL SWELLING OF NERVES SIMILAR TO CIDP**
*A.-K. Peyer Kauffmann*1, E. Wilder-Smith2; 1Neurozentrum/Luzern/CH, 2Neurologie/Bern/CH

PS2Group5-023 **COMPARISON OF HOME-BASED VERSUS HOSPITAL-BASED SPIROMETRY MEASUREMENTS IN DUCHENNE MUSCULAR DYSTROPHY**
*C. Rummey*1, T. Meier2, M. Leinonen2, S. Hasham2, T. Voit3, O.H. Mayer4, G. Buyse For The Delos Study Group5; 1/Basel/CH, 2/Pratteln/CH, 3/London/GB, 4/Philadelphia, PA/US, 5/Leuven/BE

PS2Group5-024 **AGE-RELATED DIFFERENCES IN MUSCLE MEMBRANE POLARIZATION AS ASSESSED BY VELOCITY RECOVERY CYCLES**
J. Lee1, R. Boland-Freitas2, *K. Ng*3; 1Neurology/Sydney/AU, 2Neurology/Sydney, NSW/AU, 3Neurology/St Leonards/AU

PS2Group5-025 **EFFECTIVE DIAGNOSTIC AND TREATMENT METHODS IN VASCULAR PARKINSONISM AND PARKINSON'S DISEASE: TEMPORHYTHMAL CORRECTION**
D.T. Akramova; Neurology/Tashkent City/UZ

PS2Group5-026 **KIF5A, MUTATION CAN LEAD TO SPASTIC PARAPLEGIA (SPG10); GABAARS-VESICLE TRANSPORT MOTOR INTERACTS WITH G3BP2 AND GABARAPS**
*D.-H. Seog*1, S. Kim2; 1Biochemistry/Busan/KR, 2Neurology/Busan/KR

PS2Group5-027 **VANGL2, A CORE COMPONENT OF THE WNT PCP PATHWAY CONTRIBUTES TO NEUROMUSCULAR JUNCTION FORMATION**
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PS2Group5-028 **A 3D SYSTEM FOR MECHANICAL CHARACTERIZATION OF ARTIFICIAL SKELETAL MUSCLE MICROTISSUE**
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PS2Group5-029 **POTENTIOALLY CONFOUNDING VARIABLES OF GDF-15: MITOCHONDRIA DISEASE AND OTHER NEUROLOGICAL DISEASES**
*A. Ishii*1, S. Nohara1, Z. Miyake1, N. Tozaka1, S. Okune1, H. Takeda1, H. Tsuji1, Y. Tomidokoro1, K. Nakamagoe1, K. Ishii1, M. Watanabe1, A. Tamaoka1, S. Yatsuga2, Y. Koga2; 1Neurology/Tsukuba/JP, 2Pediatrics/Kurume/JP

PS2Group5-030 **QUANTITATIVE ESTIMATES OF ULTRASOUND IMAGING IN MUSCLE PATHOLOGY**
D. Veltsista, E. Chroni; Neurology/Patras/GR

PS2Group5-031 **CIRCULATING KV1.3+ CELLS IN PATIENTS WITH SPORADIC INCLUSION BODY MYOSITIS**
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PS2Group5-032 **ULTRASOUND IMAGING OF GENIOGLOSSUS MUSCLE FUNCTION WITH CONTRAST AGENT**
O.M. Semeryak; /Lviv/UA

PS2Group5-033 **COMPUTATIONAL SPEECH ANALYSIS AS A TOOL FOR EARLY DETECTION OF BULBAR DYSFUNCTION IN ALS PATIENTS**
*B.B. Recasens*1, D.C. Lahoz2, A.G. Solà3, M.S. Martinez1, F.A. Cabrera2, J.M. Llorens4, A.B. Corberó4, M.V. Moreno5, M.A.R. Perez6; 1Neurology/Barcelona/ES, 2/Barcelona/ES, 3Physical Medicine and Rehabilitation Department/Barcelona/ES, 4Pneumology Department/Barcelona/ES, 5Endocrinology Department/Barcelona/ES, 6Neurology Department/Barcelona/ES

PS2Group5-034 **THE IMPORTANCE OF A NON-INVASIVE SCREENING IN PROXIMAL MYOPATHIES**
G. Bruno, L. Allegorico, L. Lombardi, G. Di Iorio, S. Sampaolo; Second Neurology/Naples/IT

PS2Group6-001 **CLINICAL DIVERSITY OF P/Q-TYPE CALCIUM CHANNEL ANTIBODY-ASSOCIATED PARANEOPLASTIC DISORDERS**

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PS2Group6-002 **CLINICAL FEATURES AND TREATMENT OUTCOMES OF NEUROLYMPHOMATOSIS IN KOREA**
D.H. Sung; Department of Physical and Rehabilitation Medicine/Seoul/KR

PS2Group6-003 **PEDIATRIC EXTRADURAL COMPRESSIVE MYELOPATHY SECONDARY TO GANGLIONEUROBLASTOMA; NERVE ROOTS AND BRACHIAL PLEXUS BEWARE**
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PS2Group9-001 **PSYCHOLOGICAL ASPECTS IMPACTING QUALITY OF LIFE OF PATIENTS WITH MYOPATHY**
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PS2Group9-002 **AN UNCOMMON CO-EXISTENCE OF INHERITED AND CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY**
*I. Glāzere*1, S. Šetlere2, I. Kazaine2, G. Rozentāls2; 1Neurology/Riga/LV, 2Pediatric Neurology/Riga/LV

PS2Group9-003 **ASSESSING IDEBENONE'S IMPACT ON RESPIRATORY FUNCTION IN DUCHENNE MUSCULAR DYSTROPHY: META-ANALYSIS OF TWO CLINICAL TRIALS**
*C. Rummey*1, M. Leinonen2, S. Hasham2, T. Voit3, O.H. Mayer4, G. Buyse For The Delos Study Group5; 1/Basel/CH, 2/Pratteln/CH, 3/London/GB, 4/Philadelphia, PA/US, 5/Leuven/BE

PS2Group9-004 **DEMANDS FROM THE PSYCHOLOGY OF HEALTH FOR THE CARE OF PEOPLE WITH STEINERT**
*A.J. Solernou Ferrer*1, A.L. Ruiz2, T.Z. Vaillant3, C.E. Oyola Valdizán4; 1Department of Neurogenetics/Havana/CU, 2Faculty of Psychology/Havana/CU, 3/Havana/CU, 4/Lima/PE

PS2Group9-005 **POMPHOLYX AND ECZEMATOUS SKIN REACTIONS AFTER INTRAVENOUS IMMUNOGLOBULIN THERAPY: CASE REPORTS AND REVIEW OF THE LITERATURE**
*B.-N. Yoon*1, J.-W. Yang2; 1Neurology/Marrenarero Gung-gu/KR, 2/Incheon/KR

PS2Group9-006 **GLOBAL VS INDIVIDUAL MUSCLE FATTY DEGENERATIVE CHANGES TO MONITOR DISEASE PROGRESSION IN IMMUNE-MEDIATED NECROTIZING MYOPATHY**
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PS2Group9-007 **KCNK9 IMPRINTING SYNDROME WITH CONGENITAL HYPOTONIA, DYSMORPHISM AND DEVELOPMENTAL DELAY DUE TO NOVEL MUTATION P.ALA237ASP**
*P. Seeman*1, J. Haberlova2, L. Sedláčková1, M. Šedivá1, P. Laššuthová1; 1Child Neurology, DNA Laboratory/Praha/CZ, 26 Department of Child Neurology/Prague/CZ

PS2Group9-008 **QUANTITATIVE NMR OUTCOME MEASURES FOR SKELETAL MUSCLE CHARACTERIZATION OF DUCHENNE MUSCULAR DYSTROPHY PATIENTS**
T. Gerhalter1, L.V. Gast1, B. Marty2, R. Trollmann3, S. Schüssler3, F. Roemer1, F.B. Laun1, M. Uder1, R. Schröder4, P.G. Carlier2, *A.M. Nagel*1; 1Institute of Radiology/Erlangen/DE, 2AIM & CEA NMR laboratory, Neuromuscular Investigation Center/Paris/FR, 3Department of Pediatrics/Erlangen/DE, 4Neuropathology/Erlangen/DE

PS2Group9-009 **ASSESSMENT OF LIVER SAFETY USING AN EMERGING LIVER BIOMARKER, GLUTAMATE DEHYDROGENASE**
*S. Chowdhury*1, R. Osahon2, D. Peters1, G. Layton2, A. Heatherington1, D. Roblin2, F. Muntoni3; 1/Cambridge, MA/US, 2/Abingdon/GB, 3/London/GB

PS2Group9-010 **PERCEIVED QUALITY OF LIFE AND MOTOR SCALES OUTCOMES IN TYPE 2 AND 3 SMA PATIENTS: ARE THEY RELATED?**
A.L. Frongia1, *I. Zschaeck*1, M.M.A. Alarcon Cornejo1, D. Natera De Benito1, L. Carrera-García2, J.F. Mata1, N. Padros1, O. Moya1, J. Armas1, J. Medina1, C. Ortez1, J. Colomer3, A. Nascimento1; 1Neuromuscular Disorders Unit/Barcelona/ES, 2Unidad de Patología Neuromuscular./Barcelona/ES, 3/Barcelona/ES

PS2Group9-011 **DEVELOPMENT OF A PSYCHOSOCIAL INTERVENTION PROGRAMME FOR MOTOR NEURON DISEASE**
*P.T. Thomas*1, M. Warrier1, A. Nalini2; 1Department of Psychiatric Social Work/Bangalore/IN, 2Department of Neurology/Bangalore/IN

PS2Group9-012 **HTLV-1-SPECIFIC CTLS IN THE SPINAL CORDS OF ASYMPTOMATIC HTLV-1 C ARRIERS**
E. Matsuura; Neurology and Geriatrics/Kagoshima City/JP

PS2Group9-013 **AAV-ASSOCIATED SENSORY GANGLIONITIS AND AXONOPATHY IN LABORATORY ANIMALS: A RETROSPECTIVE STUDY**
J. Hordeaux, E.L. Buza, L. Richman, C. Hinderer, N. Katz, P. Bell, J.M. Wilson; Gene Therapy Program, Department of Medicine/Philadelphia, PA/US

PS2Group9-014 **OSTEOPOROSIS IN DUCHENNE MUSCULAR DISTROPHY PATIENTS**
F.M. Sertpoyraz1, *F. Baydan*2; 1Physical Therapy and rehabilitation clinic/İzmir/TR, 2Pediatric Neurology/İzmir/TR

PS2Group9-015 **NEUROMUSCULAR GENETIC REGISTRY IN RUSSIA**
*D. Vlodavets*1, A. Monakhova1, D. Reshetov2, S. Artemyeva1, I. Shulyakova1, O. Shidlovskaya1, E. Belousova1; 1Russian Children Neuromuscular Center/Moscow/RU, 2/Moscow/RU

PS2Group9-016 **ALTERNATIVE ANALYSES OF RESPIRATORY FUNCTION IN DUCHENNE MUSCULAR DYSTROPHY: CONSISTENT TREATMENT BENEFIT OF IDEBENONE**
*O.H. Mayer*1, M. Leinonen2, C. Rummey3, T. Meier2, S. Hasham2, T. Voit4, G. Buyse For The Delos Study Group5; 1/Philadelphia, PA/US, 2/Pratteln/CH, 3/Basel/CH, 4/London/GB, 5/Leuven/BE

PS2Group9-017 **MISDIAGNOSIS OF DUCHENNE MUSCULAR DYSTROPHY**
I.D.L. Kalil, J.D.L. Fernandes, A.J. Godoy; /Alto De Pinheiros - São Paulo/BR

PS2Group9-018 **ENSURING HIGH QUALITY MUSCLE BIOPSIES AND MAGNETIC RESONANCE BIOMARKERS IN THE PHASEOUT DMD STUDY**
*J. Tinsley*1, Y. Hasoglu1, M.A. Boss2, D. Frank2, C. Faelan3, K. Vandenborne4, F. Muntoni5; 1/Abingdon/GB, 2/Cambridge, MA/US, 3/Westminster, CO/US, 4Department of Physical Therapy/Gainesville, FL/US, 5/London/GB

PS2Group9-019 **TREAT-NMD: ADVANCING DIAGNOSIS, TREATMENT AND CARE IN NEUROMUSCULAR RARE DISEASES**
A. Oyewole1, *J. Lee*1, R. Leary1, K. Bushby1, A. Aartsma-Rus2, J. Kirschner3, K. Flanigan4; 1The John Walton Muscular Dystrophy Research Centre/Newcastle/GB, 2Human Genetics/Leiden/NL, 3/Freiburg/DE, 4/Columbus/US

Monday, July 9, 2018

17:15 - 18:30 **Guided Poster Session**
Room: Mezzanine Floor Gallery and Foyers

- Topic Group 2: Diseases of Neuromuscular Junction: Clinical Features, Pathophysiology, Therapy
- Topic Group 4: Motor Neuron Diseases: Clinical Features, Pathophysiology, Therapy
- Topic Group 7: Patient Related Issues
- Topic Group 8: History

PS3Group2-001 **THYMECTOMY LOWERS THE MYASTHENIA GRAVIS BIOMARKER MIR-150-5P**
*C.J. Molin*1, A.R. Punga2, L. Sabre2, T. Punga2, C.-A. Weis3; 1Neuroscience/Uppsala/SE, 2/Uppsala/SE, 3Pathology/Mannheim/DE

PS3Group2-002 **EFFICACY OF SUBCUTANEOUS IMMUNOGLOBULIN IN MYASTHENIA GRAVIS EXACERBAION**
*Z.A. Siddiqi*1, G. Beecher2, D. Anderson3; 1Neurology/Medicine/Edmonton, /CA, 2Neurology/Medicine/Edmonton, AB/CA, 3/Edmonton, AB/CA

PS3Group2-003 **FEASIBILITY AND SAFETY OF SUBCUTANEOUS IMMUNOGLOBULIN IN MYASTHENIA GRAVIS EXACERBATION**
*Z.A. Siddiqi*1, G. Beecher2, D. Anderson3; 1Neurology/Medicine/Edmonton/CA, 2Neurology/Medicine/Edmonton, AB/CA, 3/Edmonton, AB/CA

PS3Group2-004 **A DOUBLE-BLIND PLACEBO-CONTROLLED STUDY TO EVALUATE SAFETY AND EFFICACY OF FCRN ANTAGONIST ARGX-113 IN GENERALIZED MG**
J. Verschuuren1, V. Bril2, R. Mantegazza3, A. Szczudlik4, S.R. Beydoun5, F. Rodriguez De Rivera6, M.R. Rottoli7, F. Piehl8, P. Van Damme9, T. Vu10, P. Ulrichts11, K. Verschuuren11, *A. Guglietta*11, H. De Haard11, N. Leupin11, J.F. Howard Jr12; 1/Leiden/NL, 2/Toronto/CA, 3/Milan/IT, 4/Krakow/PL, 5/Los Angeles, CA/US, 6/Madrid/ES, 7/Bergamo/IT, 8/Stockholm/SE, 9/Leuven/BE, 10/Tampa, FL/US, 11/Zwijnaarde/BE, 12/Chapel Hill, NC/US

PS3Group2-005 **WHEN IS BETTER GOOD ENOUGH? PATIENT ACCEPTABLE STATES IN MYASTHENIA GRAVIS**
*C. Barnett*1, M. Mendoza2, H. Katzberg1, V. Bril2; 1/Toronto, ON/CA, 2/Toronto/CA

PS3Group2-006 **BIOMARKER DISCOVERY IN CHILDHOOD REFRACTORY OCULAR MYASTHENIA GRAVIS**
C. Khongkhatithum, K. Srisuwan, S. Chutipongtanate, L. Thampratankul, A. Visudthibhan; Pediatrics/Bangkok/TH

PS3Group2-007 **DIFFERENCES BETWEEN THYMOMATOUS AND NON-THYMOMATOUS MYASTHENIA GRAVIS**
*F. Aguirre*1, A. Manin2, A. Villa1; 1Neurology/Buenos Aires/AR, 2Neurology/Buenos Aires City/AR

PS3Group2-008 **THYMOMA-ASSOCIATED MYASTHENIA GRAVIS IN ARGENTINA ▫**
*F. Aguirre*1, A. Manin2, V. Andres1; 1Neurology/Buenos Aires/AR, 2Neurology/Buenos Aires City/AR

PS3Group2-009 **EPIDEMIOLOGICAL FEATURE OF MYASTHENIA GRAVIS IN SOUTHERN CHILEAN POPULATION**
*M. Fuentealba*1, M. Padilla2, F. Maturana2; 1Medicine/Concepcion/CL, 2/Concepcion/CL

PS3Group2-010 **MYASTHENIA GRAVIS: THYMECTOMY IN CHILE (MGTC)**
*M. Fuentealba*1, R. Gonzalez2, A. Riquelme2; 1Medicine/Concepcion/CL, 2/Concepcion/CL

PS3Group2-011 **RESPONSE TO ECULIZUMAB IN PATIENTS WITH ACHR+ REFRACTORY MYASTHENIA GRAVIS RECENTLY TREATED WITH CHRONIC IVIG**
*S. Jacob*1, H. Murai2, K. Utsugisawa3, R.J. Nowak4, H. Wiendl5, K.P. Fujita4, F. O'Brien4, J.F. Howard Jr6; 1/Birmingham/GB, 2/Narita/JP, 3/Hanamaki/JP, 4/New Haven, CT/US, 5/Münster/DE, 6/Chapel Hill, NC/US

PS3Group2-012 **ECULIZUMAB REDUCES EXACERBATION RATES IN PATIENTS WITH ACHR+ REFRACTORY GENERALIZED MYASTHENIA GRAVIS**
*S. Jacob*1, J.T. Guptill2, A. Meisel3, K.P. Fujita4, K. Patra4, J.F. Howard Jr5; 1/Birmingham/GB, 2/Durham, NC/US, 3/Berlin/DE, 4/New Haven, CT/US, 5/Chapel Hill, NC/US

PS3Group2-013 **NONLETHAL CHRNA1-RELATED CONGENITAL MYASTHENIC SYNDROME CAUSED BY NOVEL HETEROZYGOUS MISSENSE MUTATIONS**
*D. Natera De Benito*1, L. Carrera-García2, A.L. Frongia3, L. González Quereda3, P. Gallano Petit3, C. Jou3, A. Codina3, C. Jimenez Mallebrera3, C. Ortez3, J. Colomer3, A. Nascimento3; 1Neuromuscular Disorders Unit/Barcelona/ES, 2Unidad de Patología Neuromuscular./Barcelona/ES, 3/Barcelona/ES

PS3Group2-014 **THE FIRST KOREAN CASE OF COLQ-MUTANT MYASTHENIC SYNDROME**
S.-H. Jung, J.-H. Shin, D.-S. Kim; Neurology/Kyung Sang Nam Do/KR

PS3Group2-015 **PREVALENCE AND CHARACTERISTICS OF OBSTRUCTIVE SLEEP APNEA IN KOREAN MYASTHENIA GRAVIS PATIENTS**
*J.-S. Park*1, S.-J. Heo2, J.-M. Park3, Y.J. Lee4; 1Department of Neurology/Daegu/KR, 2Department of Otorhinolayngology-Head & neck surgery/Daegu/KR, 3Department of Neurology/Gyeongju/KR, 4Pediatrics /Daegu/KR

PS3Group2-016 **PHARMACOLOGICAL TREATMENT ADHERENCE IN PATIENTS WITH MYASTHENIA GRAVIS: ASSOCIATED FACTORS AND CLINICAL CONSEQUENCES**
B.K. Vitturi, A. Pellegrinelli, B.C.O. Valério; Neurology/São Paulo/BR

PS3Group2-017 **SOCIAL CONSEQUENCES AND QUALITY OF LIFE OF PATIENTS WITH MYASTHENIA GRAVIS**
B.K. Vitturi, A. Pellegrinelli, B.C.O. Valério; Neurology/São Paulo/BR

PS3Group2-018 **HIGH EFFICACY AND SAFETY OF RITUXIMAB FOR MYASTHENIA GRAVIS: A NATIONWIDE STUDY BY AUSTRIAN ADULT NEUROLOGISTS**
*R. Topakian*1, F. Zimprich2, S. Iglseder3, N. Embacher4, M. Guger5, K. Stieglbauer6, D. Langenscheidt7, J. Rath2, S. Quasthoff8, P. Simschitz9, J. Wanschitz10, P. Müller1, D. Oel1, S. Einsiedler1, G. Schustereder1, D. Windisch11, W. Löscher10; 1Neurology/Wels/AT, 2Department of Neurology/Vienna/AT, 3Neurology/Linz/AT, 4/St. Pölten/AT, 5Department of Neurology II/Linz/AT, 6/Linz/AT, 7/Rankweil/AT, 8/Graz/AT, 9/Klagenfurt/AT, 10/Innsbruck/AT, 11/Bruck/AT

PS3Group2-019 **DIFFERENTIAL RESPONSE TO RITUXIMAB IN ACHR AND MUSK ANTIBODY POSITIVE MYASTHENIA GRAVIS: A SINGLE-CENTER RETROSPECTIVE STUDY**
T.D. Litchman1, *B. Roy*1, V. Njike2, A. Kumar1, A. Sharma1, R.J. Nowak1; 1Neurology/New Haven, CT/US, 2/Derby/US

PS3Group2-020 **OUTCOMES IN GENERALIZED MYASTHENIA GRAVIS PATIENTS IN THE NEW MILLENIUM: DATA FROM A SINGLE-CENTER RETROSPECTIVE STUDY**
M. Tomschik, J. Rath, G. Zulehner, H. Cetin, E. Hilger, A. Paul, F. Zimprich; Department of Neurology/Vienna/AT

PS3Group2-021 **B CELL TARGETED TREATMENT IN MYASTHENIA GRAVIS (BEATMG) - A PHASE 2 TRIAL OF RITUXIMAB IN MG: TOPLINE RESULTS**
*R.J. Nowak*1, C. Coffey2, J.M. Goldstein3, M. Dimachkie4, M. Benatar5, S. Huq6, B. Pearson7, J.W. Yankey2, L. Uribe2, L. Herbelin4, T.M. Burns8, K.C. O'Connor1, R. Conwit9, J.T. Kissel10, G.I. Wolfe11, D.A. Hafler1, M.E. Cudkowicz12, R.J. Barohn4, Neuronext Nn103 Beatmg Study Team; 1Neurology/New Haven, CT/US, 2Clinical Trials Statistical Data Management Center/Iowa City/US, 3Neurology/New York City/US, 4Neurology/Kansas City, KS/US, 5Neurology/Miami/US, 6/Boston/US, 7/Iowa City/US, 8Department of Neurology/Charlottesville/US, 9Neurology/Bethesda/US, 10Neurology/Columbus/US, 11Department of Neurology/Buffalo/US, 12Neurology/Boston/US

PS3Group2-022 **CONGENITAL MYASTHENIC SYNDROME: AN INDIAN SCENARIO**
*S. Balaraju*1, V.P. Kumar2, A. Töpf3, S. Vengalil2, K. Polavarapu2, S. Nashi2, A. Roos3, R. Horvath3, N. A2, H. Lochmüller4; 1John Walton Muscular Dystrophy Research Center/Newcastle/GB, 2/Bengaluru/IN, 3John Walton muscular dystrophy research center/Newcastle/GB, 4Institute of Genetic Medicine/Newcastle Upon Tyne/US

PS3Group2-023 **'MINIMAL SYMPTOM EXPRESSION' IN ACETYLCHOLINE RECEPTOR-POSITIVE REFRACTORY MYASTHENIA GRAVIS PATIENTS TREATED WITH ECULIZUMAB**
*J. Vissing*1, S. Jacob2, K.P. Fujita3, F. O'Brien3, J.F. Howard Jr4; 1/Copenhagen/DK, 2/Birmingham/GB, 3/New Haven, CT/US, 4/Chapel Hill, NC/US

PS3Group2-024 **THE PLACEBO EFFECT IN MYASTHENIA GRAVIS: A META-ANALYSIS**
*E. Frisaldi*1, A. Shaibani2, J. Vollert3, 4, B. Ferrero1, R. Carrino1, H. Ibraheem2, L. Vase5, F. Benedetti1, 6; 1Department of Neuroscience "Rita Levi Montalcini"/Turin/IT, 2Nerve & Muscle Center of Texas/Houston, TX/US, 3Center of Biomedicine and Medical Technology Mannheim CBTM/Mannheim/DE, 4Department of Surgery & Cancer, Imperial College

London/London/GB, 5Department of Psychology and Behavioural Sciences/Aarhus/DK, 6Plateau Rosa Laboratories, Breuil-Cervinia, Italy/Zermatt/CH

PS3Group2-025 **CONGENITAL MYASTHENIC SYNDROMES IN A SUBPOPULATION OF THE NORTH OF PORTUGAL**
*L. Sousa*1, E. Santos1, J. Martins2, A. Sousa3, M. Cardoso3, F. Silveira1, G. Nadais1, R. Maré4, A. Veiga5, C.S. Santos6, T. Coelho3, M. Santos7; 1Neurology/Porto/PT, 2Neurology/Matosinhos/PT, 3Neurophysiology/Porto/PT, 4Neurology/Braga/PT, 5Neurology/Vila Real/PT, 6Neurology/Santa Maria Da Feira/PT, 7Neuropediatrics/-/PT

PS3Group2-026 **FETAL ACETYLCHOLINE RECEPTOR INACTIVATION SYNDROME: A RARE, BUT POTENTIALLY TREATABLE CAUSE OF FAMILIAL MYOPATHY**
D. Nguyen1, S. Botez2, *C.-T.E. Nguyen*3; 1/Montreal, QC/CA, 2Neurology/Montreal, QC/CA, 3Pediatric Neurology/Montreal, QC/CA

PS3Group2-027 **CLINICAL AND SEROLOGICAL PREDICTORS OF THYMOMA RECURRENCES IN MYASTHENIA GRAVIS**
A. De Rosa, R. Ricciardi, M. Maestri, M. Guida, S. Rizzo, A. Chella, F. Melfi, M. Lucchi, U. Bonuccelli, G. Siciliano; /Pisa/IT

PS3Group2-028 **THE EFFECTS OF NEUROTROPHIC FACTORS ON HUMAN MUSCLE CELLS: A COMPARISON WITH MURINE MUSCLE CELLS**
S. Barbeau, F. Semprez, C. Legay; /Paris Cedex/FR

PS3Group2-029 **A PILOT STUDY OF ENGINEERED AGRIN AND ITS EFFECT ON THE SEVERITY OF EXPERIMENTAL AUTOIMMUNE MYASTHENIA GRAVIS**
Z. Li1, 2, M. Li1, 2, K. Wood1, S. Hettwer3, S. Muley4, F.-D. Shi1, 2, Q. Lui1, 2, *S. Ladha*4; 1Neurobiology/Phoenix/US, 2/Tianjin/CN, 3/Schlieren-Zurich/CH, 4Neurology/Phoenix/US

PS3Group2-030 **MUTATIONS IN GFPT1-RELATED CMS UNDERLIE A TUBULAR AGGREGATES MYOPATHY WITH SYNAPTOPATHY**
*S. Bauché*1, G. Vellieux1, D. Sternberg2, M.-J. Fontenille1, G. Brochier3, J. Messéant1, M. Fardeau3, N. Romero3, E. Fournier4, N. Streichenberger5, V. Manel5, A. Lacour6, A. Nadaj-Pakleza7, S. Sukno8, F. Bouhour9, P. Laforet4, B. Fontaine1, L. Strochlic1, B. Eymard1, F. Chevessier10, T. Stojkovic4, S. Nicole1; 1Neurogenetic and physiology/Paris/FR, 2Cardiogénétique et Myogénétique/Paris/FR, 3Unité de Pathologies neuromusculaires, Institut de Myologie/Paris/FR, 4/Paris/FR, 5/Lyon/FR, 6/Lille/FR, 7/Angers/FR, 8/Béthune/FR, 9/Bron/FR, 10/Erlangen/DE

PS3Group2-031 **THE POTENTIAL OF A NOVEL ANTIGEN-SPECIFIC GLYCOPOLYMER IN THE TREATMENT OF PATIENTS WITH MULTIFOCAL MOTOR NEUROPATHY**
*D. Demeestere*1, H. Prescher1, B. Ernst1, P. Hänggi2, R. Herrendorff2; 1Institute of Molecular Pharmacy/Basel/CH, 2R&D/Basel/CH

PS3Group2-032 **RELATIONSHIP OF ACETYLCHOLINE RECEPTOR ANTIBODY TITER AND MYASTHENIA GRAVIS SEVERITY IN CIPTO MANGUNKUSUMO HOSPITAL**
*F.T. Widayati*1, A.Y. Safri1, F. Octaviana1, L.A. Indrawati1, D. Wulandari2, T. Loho2, M. Hakim1; 1Neurology/Jakarta/ID, 2Clinical Pathology/Jakarta/ID

PS3Group2-033 **THE IMPACT OF REFRACTORY MYASTHENIA GRAVIS (MG) ON PATIENT HEALTH-RELATED QUALITY-OF-LIFE (QOL)**
A.N. Boscoe1, H. Xin2, G.J. L'Italien3, *L. Harris*3, G.R. Cutter4; 1/Cambridge, MA/US, 2/Birmingham, AL/US, 3/New Haven/US, 4Biostatistics/Birmingham, AL/US

PS3Group2-034 **VOCAL CORD PARALYSIS: A RARE PRESENTATION OF MYASTHENIA GRAVIS** ▫
V. Montes, S. Sousa, R. Guerreiro, C. Carmona, F. Pita;
Neurology/Alcabideche (Cascais) - Lisboa/PT

PS3Group2-035 **SERONEGATIVE MYASTHENIA GRAVIS WITH ANTI-LRP4 ANTIBODIES**
*J.E. Kim*1, K.H. Park2, H.S. Seo3, J.-J. Sung4, Y.-H. Hong3; 1Department of Neurology/Seoul/KR, 2Neurology/Jinju/KR, 3/Seoul/KR, 4Neurology/Seoul/KR

PS3Group2-036 **INHIBITION OF ACETYLCHOLINE RECEPTOR FUNCTION BY MYASTHENIA GRAVIS SERA DEPENDS ON RECEPTOR CLUSTERING**
*H. Cetin*1, R. Webster2, W. Liu2, I. Koneczny3, F. Zimprich1, J. Cossins2, D. Beeson2, A. Vincent2; 1Department of Neurology/Vienna/AT, 2Nuffield Department of Clinical Neurosciences/Oxford/GB, 3Institute of Neurology/Vienna/AT

PS3Group2-037 **ECULIZUMAB SHOWS CONSISTENCY OF IMPROVEMENT ACROSS MUSCLE GROUPS IN PATIENTS WITH ACHR-POSITIVE REFRACTORY MYASTHENIA GRAVIS**
*R. Mantegazza*1, K.P. Fujita2, F. O'Brien2, J.F. Howard Jr3; 1/Milan/IT, 2/New Haven, CT/US, 3/Chapel Hill, NC/US

PS3Group2-038 **ACHIEVEMENT OF MINIMAL MANIFESTATIONS IN ECULIZUMAB-TREATED ACHR-POSITIVE REFRACTORY MYASTHENIA GRAVIS PATIENTS**
*R. Mantegazza*1, G. Wolfe2, S. Muppidi3, H. Wiendl4, K.P. Fujita5, F. O'Brien5, J.F. Howard Jr6; 1/Milan/IT, 2/Buffalo, NY/US, 3/Stanford, CA/US, 4/Münster/DE, 5/New Haven, CT/US, 6/Chapel Hill, NC/US

PS3Group4-001 **INCREASED GLUCOSE FLUCTUATION IN MOTOR NEURON DISEASE DURING ENTERAL NUTRITION**
*H. Takada*1, S. Kon1, Y. Ogasawara2, T. Ota3; 1Neurology/Aomori/JP, 2Medical Technology/Aomori/JP, 3Nursing/Aomori/JP

PS3Group4-002 **PD AND SAFETY DATA FROM JEWELFISH, A STUDY OF RG7916 IN SMA PATIENTS PREVIOUSLY ENROLLED IN A SMN2-SPLICING MODIFIER STUDY**
*C.A. Chiriboga*1, E. Mercuri2, D. Fischer3, D. Kraus3, N. Thompson3, G. Armstrong4, H. Kletzl3, M. Gerber3, Y. Cleary3, T. Bergauer3, K. Gorni3, O. Khwaja3; 1Department of Neurology/New York/US, 2Paediatric Neurology and Nemo Center /Rome/IT, 3/Basel/CH, 4/Welwyn Garden City/GB

PS3Group4-003 **MLPA BASED SMN1 DELETION ANALYSIS: CLINICAL CORRELATION IN INDIAN PATIENTS WITH SPINAL MUSCULAR ATROPHY**
H. Padmanabha1, *V. Preethish-Kumar*1, K. Polavarapu1, S. Nashi2, S. Vengalil1, D. Sekar2, A. Nalini3, P. Thomas1, K. Gk1;
1Neurology/Bengaluru/IN, 2/Bengaluru/IN, 3Neurology//IN

PS3Group4-004 **ACUTE MOTOR NEURONOPATHY ASSOCIATED TO AN INFECTION BY TICK-BORNE ENCEPHALITIS VIRUS**
L. Bauer1, M. Harlay2, A. Echaniz-Laguna1, R. Herbrecht3, V. Castelain2, *J.-B. Chanson*1; 1Neurologie/Strasbourg/FR, 2Réanimation/Strasbourg/FR, 3Hématologie/Strasbourg/FR

PS3Group4-005 **ASSOCIATION OF AMYOTROPHIC LATERAL SCLEROSIS AND MULTIPLE SCLEROSIS: A CASE REPORT**
*B.K. Vitturi*1, C.P. Tilbery2, W.L. Sanvito1; 1Neurology/São Paulo/BR, 2Centro de Atendimento e Tratamento de Esclerose Múltipla (CATEM)/São Paulo/BR

PS3Group4-006 **EXPLORATION OF THE REPOSITIONING POTENTIAL OF MARKETED DRUGS FOR NEUROPATHIES**
J. Gide1, F. Roussange2, H. Polvèche1, D. Auboeuf3, A. Boland4, J.-F. Deleuze4, B. Knut5, J. Tournois1, M. Peschanski6, C. Martinat1, *S. Baghdoyan*2; 1I-STEM/Corbeil Essonnes/FR, 2Motor Neurone Disease/Corbeil-Essonnes/FR, 3Laboratoire de Biologie et Modélisation de la Cellule,/Lyon/FR, 4Centre National de Genotypage/Evry/FR, 5/Gottingen/DE, 6I-STEM CECS/Corbeil Essonnes/FR

PS3Group4-007 **DISSECTING UBA5 DYSFUNCTION IN NERVOUS SYSTEM DISORDERS USING THE ZEBRAFISH**
*R. Serrano*1, V. Oorschot2, G. Ramm2, R. Bryson-Richardson1; 1School of Biological Sciences/Melbourne, VIC/AU, 2Ramaciotti Centre for cryo-Electron Microscopy/Melbourne, VIC/AU

PS3Group4-008 **BRANAPLAM IN TYPE 1 SPINAL MUSCULAR ATROPHY: RESPIRATORY SUPPORT AND FEEDING**
*N. Deconinck*1, A. Born2, G. Baranello3, E. Bertini4, N. Goemans5, R. Pingili6, J. Praestgaard7, R. Roubenoff8, U. Schara9, E. Voltz8; 1Department Pediatric Neurology/Brussels/BE, 2/Copenhagen,/DK, 3/Milano/IT, 4Unit of Neuromuscular Disorders, Laboratory of Molecular Medicine/Rome/IT, 5/Leuven/BE, 6/East Hanover, NJ/US, 7/Novartis Pharmaceuticals, East Hanover, NJ/US, 8/Basel/CH, 9/Essen/DE

PS3Group4-009 **NUSINERSEN TREATMENT IN LONGSTANDING ADULT SMA TYPE 3 PATIENTS**
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PS3Group4-010 **DIAGNOSIS AND DIFFERENTIAL DIAGNOSIS OF SPINAL MUSCULAR ATROPHY**
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PS3Group4-011 **A CASE OF HEXANUCLEOTIDE REPEAT EXPANSION CAUSING AMYOTROPHIC LATERAL SCLEROSIS**
P.D. Shanmugarajah, J.O.S. Archer; Neurology/Wakefield/GB

PS3Group4-012 **A DE NOVO HETEROZYGOUS POINT MUTATION IN THE DYNC1H1 GENE CAUSING SPINAL MUSCULAR ATROPHY WITH LOWER EXTREMITY DOMINANT**
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PS3Group4-013 **SMARD1: A RARE CAUSE OF HYPOTONIA AND RESPIRATORY FAILURE IN INFANCY**
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PS3Group4-014 **NUSINERSEN EXPERIENCE IN INDIVIDUALS WITH SPINAL MUSCULAR ATROPHY TYPE III: A CASE SERIES**
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PS3Group4-015 **RG7916 SIGNIFICANTLY INCREASES SMN PROTEIN IN SMA TYPE 1 BABIES**
*G. Baranello*1, L. Servais2, J. Day3, N. Deconinck4, E. Mercuri5, A. klein6, B. Darras7, R. Masson1, H. Kletzl8, Y. Cleary8, G. Armstrong9, T. Seabrook8, C. Czech8, M. Gerber8, F. Lee8, K. Gelblin8, S. Nave8, K. Gorni8, O. Khwaja8; 1Developmental Neurology Unit/Milan/IT, 2/Paris/FR, 3Department of Neurology/Palo Alto, CA/US, 4/Ghent/BE, 5Paediatric Neurology and Nemo Center /Rome/IT, 6/Bern/CH, 7/Boston, MA/US, 8/Basel/CH, 9/Welwyn Garden City/GB

PS3Group4-016 **PATHOGENIC VARIANT OF THE REEP1 GENE IN A KOREAN FAMILY WITH AUTOSOMAL DOMINANT HEREDITARY SPASTIC PARAPLEGIA**
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PS3Group4-017 **NEUROFILAMENT LIGHT CHAIN AS A POTENTIAL BIOMARKER IN SPINAL MUSCULAR ATROPHY**
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PS3Group4-018 **ENHANCEMENT OF BULBAR FUNCTION IN ALS: LESSONS LEARNED FROM THE NUEDEXTA TREATMENT TRIAL**
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PS3Group4-019 **SALBUTAMOL TREATMENT IN TYPE 2 SMA PATIENTS: 18 MONTHS ASSESSMENT**
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PS3Group4-020 **VERY LATE-ONSET AMYOTROPHIC LATERAL SCLEROSIS IN A PORTUGUESE COHORT: WHICH DIFFERENCES?**
M. Oliveira Santos, M. Gromicho, S. Pinto, M. De Carvalho; /Lisbon/PT

PS3Group4-021 **ATAXIN-2 IN RUSSIAN ALS PATIENTS ATAXIN-2 IN RUSSIAN ALS PATIENTS**

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PS3Group4-022 **AVXS-101 PHASE 1 GENE THERAPY CLINICAL TRIAL IN SMA TYPE 1: EVENT-FREE SURVIVAL AND ACHIEVEMENT OF DEVELOPMENTAL MILESTONES**

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PS3Group4-023 **AN AUTOMATED ANALYSIS OF REPEATER F-WAVES IN ALS**

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PS3Group4-024 **THE OLEOS TRIAL: A LONG-TERM FOLLOW-UP OF OLESOXIME-TREATED TYPE 2 AND NON-AMBULATORY TYPE 3 SMA PATIENTS**

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PS3Group4-025 **SMA-RTER? COGNITIVE ASSESSMENT IN SPINAL MUSCULAR ATROPHY TYPE 1-2 USING EYE TRACKING**

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PS3Group4-026 **LONGER-TERM ASSESSMENT OF NUSINERSEN SAFETY/EFFICACY IN INFANTILE-ONSET SPINAL MUSCULAR ATROPHY: INTERIM ANALYSIS OF SHINE**

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PS3Group4-027 **A CASE OF KENNEDY'S DISEASE IN A PATIENT INITIALLY PRESENTED WITH RECURRENT PERIODONTITIS AND OROMANDIBULAR PAIN**

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PS3Group4-028 **UPDATED PHARMACODYNAMIC AND SAFETY DATA FROM SUNFISH PART 1, A STUDY OF ORAL RG7916 IN PATIENTS WITH TYPE 2 OR TYPE 3 SMA**

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PS3Group4-029 **MORPHINE FOR DYSPNEA IN ALS**
L. Brylev, V. Parshikov, Y. Krasnaya, E. Dikhter, E. Larin, Y. Batmanova, V. Shtabnitskiy, S. Avdeykin, A. Ataulina, D. Puzanok, A. Kasianova; /Moscow/RU

PS3Group4-030 **EXTRACELLULAR VESICLES FROM ALS SPINAL CORD AND BRAIN CONTAIN DYSREGULATED MIRNAS**
E. Feldman; /Ann Arbor/US

PS3Group7-001 **THE IMPACT OF PARTICIPATION IN CLINICAL TRIALS ON PSYCHOSOCIAL WELLBEING OF CHILDREN WITH DMD AND THEIR PARENTS**
*S. Geuens*1, J. Willen1, C. Antonis1, N. Goemans2, L. De Waele1; 1Child Neurology/Leuven/BE, 2/Leuven/BE

PS3Group7-002 **PHYSICAL ACTIVITY IN PEOPLE WITH LIMB-GIRDLE MUSCULAR DYSTROPHY AND CHARCOT-MARIE-TOOTH DISEASE IN NORWAY**
*A. Andries*1, M.R. Van Walsem1, 2, J. Frich1; 1Institute of Health and Society/Oslo/NO, 2Dept. of Neurohabilitation/Oslo/NO

PS3Group7-003 **THE JAIN FOUNDATION DYSFERLIN PATIENT REGISTRY: ACCELERATING THE PACE OF RESEARCH AND TREATMENT**
L. Rufibach, S. Shira, L. Long, B. Williams, D. Albrecht; /Seattle/US

PS3Group7-004 **PATIENT PERSPECTIVES ON THE SIDE EFFECT BURDEN OF TREATMENTS FOR MYASTHENIA GRAVIS (MG) AND THEIR IMPACT ON DAILY LIFE**
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