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19th WMS Congress – 2014 – Program

Tuesday 7 October 2014

12:30–16:00	**WMS Executive Board Meeting –** Langenbeck-Virchow-Haus
14:00–18:00	**Registration** – Langenbeck-Virchow-Haus (Foyer)
	Setting up of posters
18:30–19:30	**Opening Ceremony** – Langenbeck-Virchow-Haus (Lecture Hall) Towards an archeology of physical strength - a close look at the site of the Berlin Museum of Natural History - Hanns Zischler
20:00–23:00	**Welcome Reception** – Museum für Naturkunde, Sauriersaal

Wednesday 8 October 2014

08:30–09:00 **Congress Opening – Message from the president Victor Dubowitz** – Langenbeck-Virchow-Haus (Lecture Hall)

09:00–10:30 **Protein Aggregation, Autophagy & Proteomics;** Invited lectures (A.I.1–3) – Langenbeck-Virchow-Haus

Chairpersons: Victor Dubowitz and Werner Stenzel

A.I.1 **Basics of autophagy in muscle – cellular mechanisms**
M. Sandri

A.I.2 **The transcriptional control of autophagy in health and disease**
A. Ballabio

A.I.3 **Role of autophagy in protein aggregate and vacuolar myopathies**
C.C. Weihl

10:30–11:00 **Morning Tea and Coffee** – Langenbeck-Virchow-Haus (Foyer)

11:00–11:30 **Protein Aggregation, Autophagy & Proteomics;** Invited lectures (A.I.4) – Langenbeck-Virchow-Haus

Chairpersons: Hans-Hilmar Goebel and Montse Olivé

A.I.4 **Protein aggregate myopathies: the many faces of an expanding disease group**
R. Schröder

11:30–13:00 **Protein aggregation and disease mechanisms;** Oral presentations (G.O.1–6) – Langenbeck-Virchow-Haus

Chairpersons: Hans-Hilmar Goebel and Montse Olivé

G.O.1 **Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca^{2+} channels**
Y. Endo; S. Noguchi; Y. Hara; Y.K. Hayashi; K. Motomura; N. Murakami; S. Tanaka; S. Yamashita; R. Kizu; M. Bamba; Y. Goto; S. Miyatake; N. Matsumoto; I. Nonaka; I. Nishino

http://dx.doi.org/10.1016/j.nmd.2014.06.429

G.O.2 **Mutations in LMOD3 cause severe nemaline myopathy by disrupting thin filament organisation in skeletal muscle**
M. Kreissl; S.A. Sandaradura; J.J. Dowling; A.S. Kostyukova; N. Moroz; K.G. Quinlan; V. Lehtokari; G. Ravenscroft; E.J. Todd; O. Ceyhan-Birsoy; D.S. Gokhin; J. Maluenda; M. Lek; F. Nolent; C.T. Pappas; S.M. Novak; A. D'Amico; E. Malfatti; B.P. Thomas; S.B. Gabriel; N. Gupta; M.J. Daly; B. Ilkovski; P.J. Houweling; L.C. Swanson; C.A. Brownstein; V.A. Gupta; L. Medne; P. Shannon; A. Flisberg; E. Holmberg; P. Van den Bergh; P. Lapunzina; L.B. Waddell; D.D. Sloboda; E. Bertini; D. Chitayat; W.R. Telfer; A. Laquerrière; C.C. Gregorio; C.A.C. Ottenheijm; C.G. Bönnemann; K. Pelin; A.H. Beggs; Y.K. Hayashi; N.B. Romero; N.G. Laing; I. Nishino; C. Wallgren-Pettersson; J. Melki; V.M. Fowler; D.G. MacArthur; K.N. North; N.F. Clarke

G.O.3 **Severe congenital actin related myofibrillar myopathy**
D. Selcen

G.O.4 **The SMCHD1 mutation spectrum in FSHD2: Novel insight in clinical variability in FSHD**
T.L. van den Boogaard; R.J.L. Lemmers; P. Camano; P.J. van der Vliet; J. Balog; N. Voermans; S.J. Tapscott; A.L. de Munain; R. Tawil; B. Bakker; S.M. Maarel

G.O.5 **Mitochondrial dysfunction reveals defective poly(A) tail regulation of specific mRNAs as a primary defect in oculopharyngeal muscular dystrophy**
C. Trollet; A. Chartier; P. Klein; N. Barbezier; T. Gidaro; F. Casas; S. Carberry; P. Dowling; L. Maynadier; G. Dickson; V. Mouly; K. Ohlendieck; G. Butler-Browne; M. Simonelig

G.O.6 **Lethal disorder of mitochondrial fission caused by mutations in DNM1L**
G. Yoon; Z. Malam; T. Paton; C. Marshall; E. Hyatt; Z. Ivakine; D. Kemaladewi; C. Forge; K.S. Lee; C. Hawkins; R.D. Cohn

13:00–14:30 **Lunch** – Langenbeck-Virchow-Haus (Foyer)

14:30–16:00 **Guided poster discussion session 1: Parallel sessions** – Thaersaal/Humboldt Graduate School

OPMD + OPDM + FSH + BMD (G.P.1–16)

Facilitators: Bernard Brais and Silvère van der Maarel

G.P.1 **Refinement of diagnosis of Becker muscular dystrophy: Results of re-analysis of DNA samples**
C.S.M. Straathof; D. van Heusden; P.F. Ippel; J.G. Post; N.C. Voermans; M. de Visser; E. Brusse; J.C. van den Bergen; A.J. van der Kooi; J.J.G. Verschuuren; H.B. Ginjaar

G.P.2 **Clinical profile of pediatric patients with Becker Muscular Dystrophy (BMD)**
K.C. Shellenbarger; I. Rybalsky; J. Bange; B.L. Wong

G.P.3 **Development of a disease severity scale for Becker muscular dystrophy**
J.C. van den Bergen; B.H.A. Wokke; P. van Damme; A.J. van der Kooi; J. De Bleecker; P. De Jonghe; J.J. Verschuuren

G.P.4 **BMD patients show relative sparing of hip flexion on muscle testing and MRI**
B.H. Wokke; J.C. van den Bergen; M. Reijnierse; C.S. Rijswijk; H.E. Kan; J.J. Verschuuren

G.P.5 **Clinical profile of a 6-year old boy with Becker Muscular Dystrophy and asymptomatic rhabdomyolysis: A case report**
S. Upadhyayula; K.C. Shellenbarger; B.L. Wong

G.P.6 **Oculopharyngeal Muscular Dystrophy with PABPN1 mutation in two Chinese Singaporean families**
Y.H.J. Chai

G.P.7 **Longitudinal study of dysphagia in oculopharyngeal muscular dystrophies**
H. Arahata; M. Sakai; G. Umemoto

G.P.8 **Dramatic Improvement after Injection Augmentation in Oculopharyngodistal Myopathy**
O. Ozcan; H. Durmus; O. Tarhan; Z. Polat; F. Deymeer; Y. Parman; P. Oflazer-Serdaroglu

G.P.9 **Adult onset distal and proximal myopathy with complete ophthalmoplegia and bulbar involvement due to de novo mutation in MYH2**
M. Cabrera; R. Junckerstorff; P.J. Lamont; N.G. Laing

G.P.10 **The DUX4 promoter mouse: The next generation**
L.M. Wallace; J. Liu; S.E. Garwick-Coppens; S.M. Guckes; C. Smith; J. McBride; S.Q. Harper

G.P.11 **SMCHD1 mutations cause FSHD type 2 and act as modifiers of disease severity**
M. Larsen; W. Kress; S. Rost; N. El Hajj; C.R. Müller

G.P.12 **Molecular defects in FAT1 are associated to Facioscapulohumeral dystrophy (FSHD)**
F. Puppo; E. Dionnet; P. Gaildrat; C. Castro; R. Bernard; E. Salort-Campana; A. Shahram; I. Nishino; M. Krahn; F. Helmbacher; N. Levy; M. Bartoli

G.P.13 **Baseline characteristics of the CINRG infantile facioscapulohumeral muscular dystrophy (FSHD) cohort**
J.K. Mah; Y.W. Chen; T. Duong; A. Cnaan; Z. Sund; L.P. Morgenroth; C. McDonald; M. Tulinius; S. Sparks; R. Webster; A. Connolly; P. Karachunski; P.R. Clemens

G.P.14 **From muscular architecture to function: The involvement of FAT1 protocadherin in FSHD**
F. Puppo; M. Sebbagh; F. Helmbacher; N. Levy; M. Krahn; M. Bartoli

G.P.15 **Aerobic exercise and cognitive behavior therapy reduce fatigue and slow progression of muscle MR fatty infiltration in FSHD**
B.G. van Engelen; N. Voet; B. Janssen; G. Bleijenberg; J. Hendriks; I. de Groot; G. Padberg; A. Heerschap; A. Geurts

G.P.16 **MR-guided muscle biopsy: A novel technique for the collection of muscle biopsies**
S. Lassche; B.H. Janssen; N.C. Voermans; J.J. Futterer; B.G.M. van Engelen

Sequencing and neuromuscular studies (G.P.17–32)

Facilitators: Gina Ravenscroft and Peter Hackman

G.P.17 **TTN a challenge for next generation sequencing**
P. Hackman; A. Evilä; B. Udd

G.P.18 **Neurogenetic disease diagnostics by targeted capture and next generation sequencing**
K. Yau; R. Allcock; K. Mina; G. Ravenscroft; M. Cabrera; R. Gooding; C. Wise; P. Sivadorai; D. Trajanoski; V. Atkinson; S. Wagner; K. Nowak; R. Duff; P. Lamont; M. Davis; N. Laing

G.P.19 **Global gene expression profile in different forms of murine muscular dystrophies**
C.F. Almeida; P.C.M. Martins; P.C.G. Onofre-Oliveira; M. Vainzof

G.P.20 **A targeted next-generation sequencing panel for diagnostic use in primary myopathies**
A. Evila; B. Udd; P. Hackman

G.P.21 **Whole exome sequencing as a diagnostic tool in neuromuscular disorders**
S. Penttilä; B. Udd

G.P.22 **Utilising next-generation sequencing to determine the genetic basis of recurrent rhabdomyolysis**
R.S. Scalco; R.D.S. Pitceathly; A. Gardiner; C. Woodward; J.M. Polke; M.G. Sweeney; S.E. Olpin; R. Kirk; E. Murphy; D. Hilton-Jones; H. Jungbluth; H. Houlden; M.G. Hanna; R. Quinlivan; R.S.G. Rhabdomyolysis Study Group

G.P.23 **Diagnostic application of targeted NGS in early onset myopathies**
A. Cho; V. Vasta; B.C. Lim; J.S. Lee S.H. Eun; K.J. Kim; Y.S. Hwang; S. Hahn; J.H. Chae

G.P.24 **Cardiomyopathy in Childhood: Results from a single tertiary care center**
E. Batu; M. Pehlivanturk; G. Haliloglu; E. Utine; N. Ulgen Tekerek; F. Kara Eroglu; I. Ertugrul; G. Hizal; K. Boduroglu; H. Topaloglu; T. Coskun; H. Ozen; G. Kale; D. Alehan

G.P.25 **Low prevalence of skeletal muscle involvement in patients suffering from idiopathic cardiomyopathy**
L. González Mera; J. Salazar; J. Roca; L. Gonzalez Quereda; P. Gallano; F. Martínez; J. González Costelo; M. Olive

G.P.26 **Vitamin D deficiency myopathy in Egypt: A treatable myopathy**
N.A. Fahmy; D. Fayez

G.P.27 **Latest Updates to the MDA Monoclonal Antibody Resource for Neuromuscular Disorders**
L.T. Le; T.M. Nguyen; C.A. Sewry; G.E. Morris

G.P.28 **Through myotubes normalization, CYTOO 2D+ increase sensitivity of muscle damage HCS assay**
Y.M. Margaron; M. Fernandes; D. Morales; S. Degot

G.P.29 **Implementing robustness of preclinical efficacy studies: Need for a concerted effort**
R. Willmann

G.P.30 **Total energy expenditure estimation can be improved by adjustments for body composition, mechanical ventilation and consciousness disturbance in patients with neuromuscular disorders**
T. Matsumura; T. Saito; H. Fujimura; S. Sakoda

G.P.31 **Voluntary running wheel activity and body weight analyses in diverse mouse strains: A platform for identifying modifying genes for neuromuscular diseases**
A.M. Messineo; J. Ma; J. Boutilier; E.L. McNamara; R. Ong; A. Wallace; G. Manship; R. Ram; M. Mehta; N.G. Laing; G. Morahan; K.J. Nowak

G.P.32 **Enhancement of myosin heavy chain class I (MyHC I) mRNA expression in C2C12 myocyte by chlorogenic acid**
J. Yamaji; Y. Mori; R. Hiroshima; M. Watanabe; A. Miyazaki

Congenital myopathies (G.P.33–50)

Facilitators: Ana Ferreiro and James Dowling

G.P.33 **Molecular analysis of a Brazilian cohort of myotubular and centronuclear myopathy patients**
O. Abath Neto; C.A. Martins; U.C. Reed; V. Biancalana; C. Bönnemann; J. Laporte; E. Zanoteli

G.P.34 **The expanding spectrum of congenital titinopathies**
C. Chauveau; C.G. Bönnemann; C. Julien; A.L. Kho; H. Marks; B. Talim; P. Maury; E. Uro-Coste; A. Alexandrovich; A. Vihola; A.R. Foley; M. Santi; B. Udd; H. Topaloglu; S.A. Moore; M. Gotthardt; M.E. Samuels; M. Gautel; A. Ferreiro

G.P.35 **Analysis of a large patient cohort with recessive truncating TTN mutations reveals novel clinical features and a diverse range of muscle pathologies**
E.C. Oates; K.S. Yau; A. Charlton; S. Brammah; M.A. Farrar; H. Sampai; P.L. Lamont; D. Mowat; R.B. Fitzsimons; A. Corbett; M.M. Ryan; H.L. Teoh; G.L. O'Grady; R. Ghaoui; S. Kaur; M. Lek; K.N. North; D.G. MacArthur; M.R. Davis; N.G. Laing; N.F. Clarke

G.P.36 **Disease specific expression patterns of skeletal muscle foetal myosin heavy chain isoform (nMHC) in congenital myopathies**
E. Rivas; R. Phadke; M. Ellis; D. Chambers; L. Feng; C. Sewry

G.P.37 **Clinical overlap between centronuclear myopathy and axonal neuropathy (CMT2) due to mutation in DNM2 gene**
J. Haberlova; D. Safka-Brozkova; M. Gonzales; S. Zuchner; P. Seeman

G.P.38 **Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations**
J. Bohm; V. Biancalana; E. Malfatti; A.L. Taratuto; M. Olive; B. Eymard; J. Weis; N.B. Romero; J. Laporte

G.P.39 **An international prospective, longitudinal study of the natural history and functional status of patients with myotubular myopathy**
M. Annoussamy; H. Landy; D. Ramsdell; M. Nelken; F. Muntoni; C. Bönnemann; D. Bharucha; J.J. Dowling; K. Amburgey; C. Lilien; G. Ollivier; J. Laporte; V. Biancalana; U. Schara; J.M. Cuisset; A. D'Amico; N. Deconinck; P.Y. Jeannet; A. Klein; J. Fluss; M. Mayer; A.M. Seferian; A.G. Le Moing; T. Gidaro; J.Y. Hogrel; F. Mingozzi; A. Buj-Bello; T. Voit; L. Servais

G.P.40 **Skeletal muscle MRI in an X-linked myotubular myopathy patient who became ambulatory**
M. Shichiji; K. Ishigaki; T. Murakami; T. Sato; S. Kajino; M. Osawa; S. Nagata; K. Saito

G.P.41 **Canine X-linked myotubular myopathy: A dose-finding study of systemic AAV8-MTM1 effects on muscle strength, gait, respiration, neuromuscular function and survival**
D.L. Mack; M. Goddard; J.M. Snyder; J. Doering; M.W. Lawlor; P. Moullier; M. O'Callaghan; A.H. Beggs; F. Mavilio; K. Poulard; V. Latournerie; A. Buj-Bello; M. Childers

G.P.42 **Mild clinical phenotype in two siblings carrying myotubular myopathy**
A. Magot; V. Biancalana; S. Mercier; A. David; G. Fayet; J.M. Mussini; J. Laporte; Y. Pereon

G.P.43 **Gene replacement therapy of myotubular myopathy: Restricting expression of MTM1 in skeletal muscle**
L. Buscara; R. Joubert; C. Moal; K. Poulard; S. Martin; A. Vignaud; F. Mavilio; A. Buj-Bello

G.P.44 **Clinical features of eight French patients with STIM 1 gene mutations**
B. Eymard; K. Ghorab; P. Laforet; F. Chevessier; J. Vallat; D. Hantai; N. Romero; J. Böhm; J. Laporte

G.P.45 **Therapy of the dynamin 2-linked centronuclear myopathy by mRNA repair using Spliceosome-mediated RNA trans-splicing (SMarT) strategy**
D. Trochet; B. Prudhon; A. Jollet; M. Bitoun

G.P.46 **Identifying new therapies for RYR1-related myopathies using a novel drug discovery pipeline**
J. Volpatti; A. Burns; A.E. Davidson; P. Roy; J.J. Dowling

G.P.47 **Severe congenital myopathy with central nuclei and novel *RYR1* gene mutations**
N. Chrestian; J. Dowling; K. Amburgey; T. Moraes; R. Cohn; C. Hawkins; W. Halliday; L. McAdam; D. Biggar; J. Vajsar

G.P.48 **Genotype-phenotype correlations in recessive *RYR1*-related myopathies**
K. Amburgey; A. Bailey; J.H. Hwang; M.A. Tarnopolsky; C.G. Bönnemann; L. Medne; K.D. Mathews; J. Collins; J.R. Daube; G.P. Wellman; B. Callaghan; J. Vajsar; G. Yoon; R. Cohn; N.F. Clarke; J.J. Dowling

G.P.49 **RYR1 mutations in adults with acute rhabdomyolysis episodes**
F. Bompaire; N. Roux-Buisson; N. Monnier; F. Feillet; N.B. Romero; P. Laforêt; A. Behin

G.P.50 **Mapping a cardiac actin expression QTL using recombinant inbred mouse models**
J. Boutilier; R. Ram; M. Mehta; Q. Thien; E.L. McNamara; R. Ong; A.M. Messineo; L. Balmer; A. Wallace; G. Manship; N.G. Laing; G. Morahan; K.J. Nowak

Distal myopathies; (G.P.51–59)

Facilitators: Stella Mitrani-Rosenbaum and Nigel Clarke

G.P.51 **Clinical and genetic characterization of distal myopathies**
M. Cabrera; R. Junckerstorff; M. Needham; P.J. Lamont; N.G. Laing

G.P.52 **Cardiac impairment in GNE myopathy**
M.C. Malicdan; L. Mian; P. Leoyklang; F. Celeste; D. Despres; P. Zerfas; N. Carrillo-Carrasco; W.A. Gahl

G.P.53 **KL-6/MUC1 is a novel diagnostic marker for GNE-myopathy**
T. Kurashige; T. Takahashi; Y. Nagano; K. Sugie; C. Watanabe; H. Maruyama; S. Ueno; M. Matsumoto

G.P.54 **In vivo GNE Interactions**
A. Harazi; M. Becker Cohen; S. Hinderlich; S. Mitrani-Rosenbaum

G.P.55 **GNE myopathy: Assessment of an adeno associated virus AAV2/8 based GNE gene delivery system**
S. Mitrani-Rosenbaum; L. Yakovlev; M. Becker Cohen; O. Rivni; A. Harazi; S. Noguchi; I. Nishino; Y. Fellig; Z. Argov

G.P.56 **A de novo mutation of the MYH7 gene in a large Chinese family with autosomal dominant myopathy**
T. Toda; H. Xiong; T. Oda; K. Kobayashi; S. Wang; W. Satake; H. Jiao; Y. Yang; Y. Suzuki; S. Sugano; X. Wu

G.P.57 **Novel MYH7-mutations in Norwegian patients with distal myopathy**
P.S. Sanaker; T. Leren; H. Miletic; L.A. Bindoff

G.P.58 **The first Asian family having vocal cord and pharyngeal weakness with distal myopathy due to a MATR3 mutation.**
T. Nishikami; S. Yamashita; N. Tawara; A. Mori; H. Hori; T. Hirahara; Y. Maeda; Y. Ando

G.P.59 **Muscle pathology as a diagnostic clue to Allgrove syndrome**
J. Reimann; N. Kohlschmidt; K. Kappes-Horn; M. Stepien-Mehring; K. Tolksdorf; K. Kuchelmeister

Inflammatory myopathies; (G.P.60–81)

Facilitators: Olivier Benveniste and Jan De Bleecker

G.P.60 **Cytoplasmic 5'-nucleotidase 1A are aggregated in type 2 fiber in sporadic inclusion body myositis**
N. Tawara; S. Yamashita; T. Nishikami; K. Kawakami; A. Tawara; H. Hori; T. Hirahara; Y. Maeda; Y. Ando

G.P.61 **Analyses of the pathogenesis in muscle-specific wild-type TDP-43 transgenic mice**
S. Yamashita; K. Kawakami; T. Nishikami; N. Tawara; Y. Maeda; Y. Ando

G.P.62 **Molecular cell stress mechanisms in an in vitro model of IBM**
K. Schmidt; M. Wienken; C. Keller; J. Schmidt

G.P.63 **Identification of widespread alterations of RNA metabolism in sporadic inclusion-body myositis using SILAC-based quantitative proteomics**
M. Berger; T. Opialla; S. Kempa; S. Spuler

G.P.64 **Proteomic analysis of rimmed vacuoles in sporadic inclusion body myositis (sIBM)**
K. Krause; A.K. Güttsches; A. Maerkens; S. Brady; M. Tegenthoff; J. Holton; K. Marcus; M. Vorgerd; R.A. Kley

G.P.65 **Obstructive sleep apnoea and subclinical impairment of respiratory function are common in sporadic inclusion body myositis**
P.M. Rodriguez Cruz; M. Needham; P. Hollingsworth; F.L. Mastaglia; D.R. Hillman

G.P.66 **Real-time MRI for the evaluation of dysphagia in inclusion body myositis (IBM)**
P.O. Carstens; S. Zhang; A. Olthoff; E. Bremen; J. Lotz; J. Frahm; J. Schmidt

G.P.67 **Myositis with invasion of endomysial cell infiltrate but without rimmed vacuoles: Separate phenotype or variant of sporadic inclusion body myositis?**
M. de Visser; J. van de Vlekkert; J.E. Hoogendijk

G.P.68 **The utility of anti-cN1A autoantibody for the diagnosis of sporadic inclusion body myositis**
H. Nihimura; S. Suzuki; S. Noguchi; A. Uruha; S. Mitsuhashi; Y.K. Hayashi; I. Nonaka; I. Nishino

G.P.69 **Th2-M2 immunity in granulomas of neuromuscular sarcoidosis and macrophagic myofasciitis**
C. Preuße; H. Goebel; D. Pehl; J.L. Rinnenthal; Y. Allenbach; F.L. Heppner; R.A. Kley; M. Vorgerd; F.J. Authier; R. Gherardi; W. Stenzel

G.P.70 **Classification of inflammatory myopathies- new avenues and future perspectives**
Y. Allenbach; O. Benveniste; C. Preusse; D. Pehl; H. Goebel; W. Stenzel

G.P.71 **Inflammatory myopathy associated with anti-MDA5 antibodies: A new entity?**
Y. Allenbach; G. Leroux; A. Rigolet; B. Hervier; T. Maisonobe; F.J. Authier; J. Aouizerate; N. Limal; A. Meyer; P. Hufnagl; N. Zerbe; C. Preusse; K. Mariampillai; S. Herson; O. Benveniste; W. Stenzel

G.P.72 **Necrotizing auto-immune Myopathies: New myopathological aspects**
Y. Allenbach; A. Rigolet; T. Stojkovic; A. Behin; B. Eymard; P. Laforet; K. Mariampillai; N. Zerbe; P. Hufnagl; C. Preusse; T. Maisonobe; S. Herson; H.H. Goebel; O. Benveniste; W. Stenzel

G.P.73 **Necrotizing autoimmune myopathy: Clinical and histopathological features of 6 patients**
H. Karasoy; A.N. Yuceyar; O. Ekmekci; M. Celebisoy

G.P.74 **Anti-Signal Recognition Particle associated myopathy in a multiethnic Malaysian population**
T. Ambang; J.S. Tan; T.E. Cheah; N. Shahrizaila; K.T. Wong; K.J. Goh

G.P.75 **A rare inflammatory myopathy with cytochrome oxidase negative muscle fiber patient which presents in chidhood: Case report**
G. Diniz; O. Yavascan; Z. Yildirim; B. Sarkis; C. Alparslan; C. Ozturk

G.P.76 **Juvenile dermatomyositis involving large muscle infarction in three cases**
R. Koichihara H. Komaki; A. Ishiyama; Y.K. Hayashi; R.S. Tsuburaya; T. Saito; Y. Saito; E. Nakagawa; K. Sugai; M. Sasaki; I. Nonaka; I. Nishino

G.P.77 **The Lymphoid Follicle Variant of juvenile Dermatomyositis**
J. Radke; D. Pehl; E. Aronica; D. Schonenberg-Meinema; U. Schneider; F.L. Heppner; M. de Visser; H.H. Goebel; W. Stenzel

G.P.78 **Acute Myositis due to Sarcocystis nesbitii infection**
K.J. Goh; K.T. Wong; N. Ramli; C.T. Tan

G.P.79 **An analysis of the sensitivity and specificity of MHC-I and MHC-II immunohistochemical staining in muscle biopsies for the diagnosis of inflammatory myopathies**
P.M. Rodriguez Cruz; L. Yue-Bei; J. Miller; R.C. Junckerstorff; V. Fabian; F.L. Mastaglia

G.P.80 **Unusual association of juvenile polyarthritis and granulomatous myositis in three siblings from a moroccan consanguineous family**
T. Gidaro; E. Bianchi; O. Hougrand; B. Florkrin; J.P. Misson; F. Barache; L. Servais

G.P.81 **A clinical point of view diagnosing juvenile idiopathic inflammatory myopathies without skin manifestations**
H. Komaki; Y. Endo; A. Ishiyama; I. Nonaka; Y.K. Hayashi; I. Nishino

DMD 1 experimental and animal approaches; (G.P.82–96)

Facilitators: Jeffrey Chamberlain and Benedikt Schoser

G.P.82 **Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle**
M.C. Walter; N. Klymiuk; A. Blutke; A. Graf; S. Krause; A. Wuensch; S. Krebs; B. Kessler; V. Zakhartchenko; M. Kurome; E. Kemter; H. Nagashima; B. Schoser; N. Herbach; H. Blum; R. Wanke; A. Aartsma-Rus; H. Lochmuller; E. Wolf

G.P.83 **Assessment of outcome measures for dystrophin deficient mice**
J. Zschuentzsch; Y. Zhang; F. Klinker; P. Jouvenal; M. Tiburcy; D. Liebetanz; H. Brinkmeier; J. Schmidt

G.P.84 **Muscle physiology properties of mouse models for Duchenne muscular dystrophy**
M. van Putten; R. Terry; M. Hulsker; K.E. Wells; A. Aartsma-Rus; D.J. Wells

G.P.85 **Truncated dystrophin with exon 45–55 deletion induced muscle atrophy and fiber type change through the hyper-nitrosylation of the ryanodine receptor type-1 and constant release of Ca^{2+} to the cytosol**
J. Tanihata; T. Nagata; T. Saito; N. Ito; Y. Aoki; A. Nakamura; S. Takeda

G.P.86 **Digital droplet RT-PCR for the absolute quantification of exon skipping induced by antisense oligonucleotides in (pre-)clinical development for Duchenne muscular dystrophy**
I.G.M. Kolfschoten; R.C. Verheul; J.C.T. van Deutekom; N.A. Datson

G.P.87 **Prospects for single antisense oligonucleotide-induced multiple exon skipping for rare non-hotspot mutations in Duchenne muscular dystrophy**
J.A.M. Janson; I.G.M. Kolfschoten; R.E.Y. van den Eijnde; R. Weij; R.C. Verheul; A. Baghat; M.M. Plug; P.C. de visser; J.C.T. van Deutekom

G.P.88 **Novel inhibitor of hematopoietic prostaglandin D synthase improves the muscle disorder in an experimental model of Duchenne muscular dystrophy**
K. Tanaka; K. Aritake; M. Tayama; E. Sasaki; T. Utsugi; T. Sasaoka; Y. Urade

G.P.89 **New orally available compounds which modulate utrophin expression for the therapy of Duchenne muscular dystrophy (DMD)**
R.J. Fairclough; S. Guiraud; S.E. Squire; A. Babbs; B. Edward; N. Shah; A. Bracchi; F.X. Wilson; G. Horne; N. Robinson; N. Araujo; D.S. Hewings; A. Vuorinen; S.G. Davies; G.M. Wynne; A.J. Russell; J. Tinsley; K.E. Davies

G.P.90 **Effects of S48168/Arm210, a new rycal® compound, on pathology related signs of exercised dystrophic mdx mouse**
R.F. Capogrosso; A. Cozzoli; A. Giustino; P. Mantuano; A.M. Massari; E. Conte; M. Cannone; M. De Bellis; A. Liantonio; G.M. Camerino; A. De Luca

G.P.91 **Diapocynin, a putative NADPH oxidase inhibitor, ameliorates the phenotype of a mouse model of Duchenne muscular dystrophy**
H.M. Ismail; L. Scapozza; U.T. Ruegg; O. Dorchies

G.P.92 **Persistence and stability at 6 months of AAV genome and dystrophin expression in GRMD dogs after forelimb delivery of a recombinant AAV carrying an exon skipping sequence**
L. Servais; C. Le Guiner; M. Montus; Y. Cherel; V. Francois; J.L. Thibaud; C. Wary; B. Matot; T. Larche; L. Guigand; M. Dutilleul; H. Goubin; J.Y. Deschamps; C. Domenger; M. Allais; J. Le Duff; M. Devaux; N. Jaulin; M. Guilbaud; O. Adjali; V. Latournerie; P. Veron; S. Boutin; C. Leborgne; D. Desgue; S. Moullec; F. Barnay-Toutain; C. Riviere; N. Delaunay; G. Bonne; C. Masurier; J.Y. Hogrel; P. Carlier; P. Moullier; T. Voit

G.P.93 **Entry and intracellular trafficking of adeno-associated viral 8 (AAV8) vector in DMD muscular cells**
B. Maniangou; B. Cadot; M. Dorna; I. Holtzman; M. Pontoriero; A. Jollet; K. Mamchaoui; S. Lorain; T. Voit; F. Pietri-Rouxel; S. Benkhelifa-Ziyyat

G.P.94 **Induction of the N-truncated dystrophin by out-of-frame exon 2 skipping restores muscle function in the Dup2 mouse, providing further support for a therapeutic pathway for 5' DMD mutations**
N. Wein; A. Vulin; T. Simmons; K.N. Heller; A. Rutherford; L.R. Rodino-Kaplac; D. Johnson; R.B. Weiss; F. Muntoni; K.M. Flanigan

G.P.95 **Optimization of micro-dystrophin cassettes for AAV-mediated gene therapy**
J.S. Chamberlain; J. Ramos; G. Banks; S.D. Hauschka

G.P.96 **Dose escalation studies of rAAV9 U7snRNA targeting exon 2 show highly efficient skipping in the Dup2 mouse**
T. Simmons; N. Wein; A. Vulin-Chaffiol; K. Heller; A. Rutherford; K. Shontz; K. Flanigan

16:00–17:30 **Guided poster discussion session 2: Parallel sessions** – Thaersaal/Humboldt Graduate School

Afternoon Tea and Coffee

DMD 2 Therapeutic evaluations and approaches; (G.P.97–116)

Facilitators: Urs Ruegg and Shinichi Takeda

G.P.97 **Prophylactic oral bisphosphonate therapy in Duchenne muscular dystrophy: The Newcastle upon Tyne experience**
A. Sarkozy; R. Srinivasan; D. Rawlings; M. Guglieri; C. Owen; V. Straub; T. Cheetham; K. Bushby

G.P.98 **Changes in height and age adjusted DXA bone indices with oral bisphosphonate treatment in Duchenne muscular dystrophy**
C. Tian; M. Rutter; L. Hornung; J. Khoury; L. Miller; J. Bange; I. Rybalsky; B. Wong

G.P.99 **The Wilmington Robotic Exoskeleton (WREX) Improves Upper Extremity Function in Patients with Duchenne Muscular Dystrophy**
T. Estilow; A. Glanzman; J. Flickinger; K.M. Powers; A. Moll; L. Medne; G. Tennekoon; S.W. Yum

G.P.100 **Pilot study of flavocoxid in ambulant DMD patients**
S. Messina; G.L. Vita; N. Licata; M. Sframeli; A. Bitto; M.G. Distefano; C. Barcellona; M. La Rosa; S. Romeo; A. Ciranni; M. Aguennouz; F. Squadrito; G. Vita

G.P.101 **CAT-1004, a novel anti-Inflammatory agent under development for treatment of Duchenne Muscular Dystrophy**
J. Milne; J. Donovan; L. Sweeney; M. Sleeper; D. Hammers; M. Jirousek; M. Curtis

G.P.102 **Utrophin modulators to treat Duchenne muscular dystrophy (DMD): Results from the Phase 1b safety and PK study in DMD boys dosed with SMT C1100**
F. Muntoni; S. Spinty; H. Roper; I. Hughes; V. Ricotti; A. Bracchi; G. Horne; J. Tinsley

G.P.103 **Biomarker development to support the clinical development of utrophin modulators for Duchenne muscular dystrophy therapy**
J. Tinsley; N. Janghra; F. Wilson; C. Sewry; G. Horne; J. Morgan

G.P.104 **Oral small molecule compounds that promote the skipping of exon 51 in the DMD gene**
N.A. Naryshkin; A. Dakka; V. Gabbeta; J. Pichardo; M.G. Woll; N. Zhang; G.M. Karp; E.M. Welch

G.P.105 **Preclinical evaluation of tamoxifen and other selective estrogen receptor modulators in mdx^{5Cv} dystrophic mice**
O.M. Dorchies; H.M. Ismail; S. Tardy; J. Reutenauer-Patte; E. Dahmane; L.A. Décosterd; D. Picard; U.T. Ruegg; L. Scapozza

G.P.106 **Internal consistency and robustness of clinical efficacy data from the phase 2b study of ataluren (PTC124®) in nonsense mutation Duchenne muscular dystrophy**
G.L. Elfring; A. Reha; R. Spiegel; S.W. Peltz; C. McDonald

G.P.107 **Complementary human skin models as a tool to study oligonucleotide-induced injection site reactions**
C. den Besten; T. Steevels; P. Ekhart; S. Jones; S. Gibbs; G. Campion

G.P.108 **Transient proteinuria with oligonucleotide therapy due to interference with tubular protein reabsorption**
C. den Besten; S. Jones; M. Wilmer; R. Masereeuw; G. Campion

G.P.109 **Evaluation of exon skipping activity of 2'-deoxy-2'-fluoro antisense oligonucleotides for Duchenne muscular dystrophy**
S.M.G. Jirka; J.W. van der Meulen; C.L. Tanganyika-de Winter; M. van Putten; M. Hiller; R. Vermue; P.C. de Visser; A. Aartsma-Rus

G.P.110 **Safety and pharmacokinetic profile of eteplirsen, SRP-4045, and SRP-4053, three phosphorodiamidate morpholino oligomers (PMOs) for the treatment of patients with Duchenne muscular dystrophy (DMD)**
P. Sazani; T. Magee; J.S. Charleston; C. Shanks; J. Zhang; M. Carver; L. Rodino-Klapac; Z. Sahenk; K. Roush; L. Bird; L.P. Lowes; L. Alfano; A.M. Gomez; S. Lewis; V. Malik; K. Shontz; K. Flanigan; C. Shilling; J. Bhalli; H. Kaur; J. Walisser; J. Forget; J. Saoud; J.R. Mendell; E. Kaye

G.P.111 **Development of an ultrasensitive ELISA method for the determination of phosphorodiamidate morpholino oligonucleotide (PMO) levels in biological samples**
U. Burki; A. Blain; S. Laval; V. Straub

G.P.112 **Pulmonary function is stable through week 120 in patients with Duchenne muscular dystrophy (DMD) treated with exon-skipping drug eteplirsen in phase 2b study**
J.R. Mendell; L.P. Lowes; L. Alfano; J. Saoud; P. Duda; E. Kaye

G.P.113 **Drisapersen (DRIS) treatment for Duchenne muscular dystrophy (DMD): Results of up to 188 weeks' follow-up of an open-label extension study**
N. Goemans; M. Tulinius; R. Wilson; C. Wardell; P. Bedwell; G. Campion

G.P.114 **Evaluation of efficacy and safety baseline parameters in patients with Duchenne muscular dystrophy (DMD) from three placebo-controlled studies of drisapersen (DRIS)**
E. Mercuri; T. Voit; N. Goemans; C.M. McDonald; R. Wilson; C. Wardell; G. Campion

G.P.115 **Pooled analyses of efficacy parameters in patients with Duchenne muscular dystrophy (DMD): Results from the drisapersen (DRIS) clinical trial programme**
N. Goemans; T. Voit; C.M. McDonald; E. Mercuri; R. Wilson; C. Wardell; G. Campion

G.P.116 **SCOPE – DMD, an EU FP7 funded consortium for skipping trail across Europe in Duchenne muscular dystrophy**
O. Veldhuizen; G. Campion; A. Morgan; H. Aygun; S. Wojczewski; T. Voit; P. Carlier; J. Verschuuren; A. Aartsma – Rus; V. Straub

Myofibrillar myopathies and autophagy; (A.P.1–16)

Facilitators: Heinz Jungbluth and Duygu Selcen

A.P.1 **Proteomic analysis in 72 myofibrillar myopathy (MFM) patients identifies new disease-relevant proteins accumulating in aggregates and reveals subtype-specific proteomic profiles**
A. Maerkens; M. Olivé; G. Tasca; K. Claeys; R. Barresi; A. Sarkozy; G. Pfeffer; T. Evangelista; S. Feldkirchner; J. Reimann; F. Hanisch; W. Stenzel; J. Schessl; B. Schoser; L. Goldfarb; B. Udd; P. Chinnery; H. Lochmüller; R. Schröder; K. Marcus; M. Vorgerd; R.A. Kley

A.P.2 **Proteomic profile of cytoplasmic bodies (CB) compared to non-CB aggregates in HMERF associated with mutations in A-band titin**
A. Maerkens; G. Tasca; G. Pfeffer; A. Sarkozy; J. Uszkoreit; R. Barresi; M. Vorgerd; B. Udd; R. Schröder; K. Marcus; H. Lochmüller; P. Chinnery; R.A. Kley

A.P.3 **Unusual extraskeletal involvement and a novel BAG3 mutation revealed by NGS screening in a large cohort of myofibrillar myopathies**
A.L. Semmler; S. Sacconi; J.E. Bach; C. Liebe; J. Bürmann; R.A. Kley; A. Ferbert; R. Anderheiden; P. Van den Bergh; J.J. Martin; P. De Jonghe; E. Neuen-Jacob; O. Müller; M. Deschauer; M. Bergmann; J.M. Schröder; M. Vorgerd; J.B. Schulz; J. Weis; W. Kress; K.G. Claeys

A.P.4 **Cytoplasmic bodies in the muscle of HMERF patients with TTN A150/FN3 119 and kinase domain mutations – An immunofluorescent analysis**
A. Vihola; J. Palmio; G. Tasca; B. Eymard; A. Evila; S. Lange; F. Xiang; L. Edstrom; P. Hackman; M. Gautel; B. Udd

A.P.5 **Novel recessive myotilin mutation cause severe myofibrillar myopathy**
J. Schessl; E. Bach; S. Rost; S. Feldkirchner; C. Kubny; S. Müller; F.G. Hanisch; W. Kress; B. Schoser

A.P.6 **Autosomal recessive myofibrillar myopathy caused by ACTA1 mutations**
M. Guglieri; N. Sambuughin; A. Sarkozy; R. Barresi; H. Lochmüller; K. Bushby; L.G. Goldfarb; V. Straub

A.P.7 **Misfolding of fibronectin III 119 subdomain in titin results in hereditary myopathy with early respiratory failure**
C. Hedberg; A. Gomez Toledo; C.M. Gustafsson; G. Larson; A. Oldfors; B. Macao

A.P.8 **Necklace cytoplasmic bodies in hereditary myopathy with early respiratory failure (HMERF)**
A. Uruha; Y.K. Hayashi; S. Mitsuhashi; S. Noguchi; I. Nonaka; I. Nishino

A.P.9 **Elucidation of the mechanism of disease in BAG3-related myofibrillar myopathy**
A.A. Ruparelia; R. Vaz; R.J. Bryson-Richardson

A.P.10 **Investigating a novel knock-in mouse model with a mutation (E247K) in the skeletal muscle-specific filamin C gene**
E.L. McNamara; G. Ravenscroft; R.M. Duff; P.B. Daniel; S.P. Robertson; N.G. Laing; K.J. Nowak

A.P.11 **The pathological spectrum of EPG5-related multisystem disorders**
S. Byrne; I. Bodi; C. Sewry; H.H. Goebel; H. Lidov; C. Dionisi-Vici; R.C. Rogers; D. Manchester; M. Al-Owain; E. Said; D. Pilz; M. Ryan; C. Marques Lourence; A.Y. Manzur; S.A. Robb; T. Cullup; T. Whyte; F. Muntoni; M. Gautel; H. Jungbluth

A.P.12 **A novel, EPG5-related vacuolar myopathy**
T. Whyte; S. Byrne; A.L. Kho; T. Cullup; S. Robb; C. Sewry; I. Bodi; N. Hart; R. Howard; M. Gautel; F. Muntoni; H. Jungbluth

A.P.13 **Myotonic Discharges in Autophagic Vacuolar Myopathies: A Potential Electrophysiologic Marker**
L.U. Gutmann; L.A. Gutmann; S.A. Moore

A.P.14 **A new in/del in the critical splicing region of the VMA21 gene causing X-linked myopathy with excessive autophagy (XMEA)**
M. Vainzof; M. Lazar; G.L. Yamamoto; C.F. Almeida; P. Onofre-Oliveira; L. Nogueira; L.U. Yamamoto; M. Zatz; H.C.A. Silva

A.P.15 **Severe early onset cardiomyopathy in females with Danon disease is not caused by skewed X-chromosome inactivation**
G. Máthé; C. Hedberg; K. Thomson; K. Karason; I. Östman-Smith A. Oldfors

A.P.16 **LysoPlex: A "preferential exome" strategy to clarify molecular mechanisms of autophagic vacuolar myopathies**
G. Di Fruscio; M. Savarese; R. De Cegli; M. Mutarelli; V. Singhmarwah; D. Di Bernardo; S. Banfi; A. Ballabio; V. Nigro

MRI; (G.P.117–130)

Facilitators: Pierre Carlier and Giuseppe Vita

G.P.117 **NMR based biomarkers to study age-related degenerative changes in the human quadriceps**
N. Azzabou; J.Y. Hogrel; P.G. Carlier

G.P.118 **Net muscle volumetry by MRI and bioelectrical impedance analysis for healthy volunteers; an observation study**
T. Nakayama; T. Uchiyama; S. Kuru

G.P.119 **Skeletal muscle fatty degenerative changes can be evaluated both qualitatively and quantitatively from whole-body Dixon NMR images with an important gain in acquisition time**
B. Marty; P.Y. Baudin; B. Robert; A. Shukelovich; N. Azzabou; P.G. Carlier

G.P.120 **The Sodium/Hydrogen Exchanger (NHE-1): A promising novel target for DMD**
U. Burki; E. Greally; S. Laval; S. Schäfer; V. Straub

G.P.121 **Fast, precise, interactive segmentation of skeletal muscle NMR images**
P.G. Carlier; A. Shukelovich; P.Y. Baudin; J.M. Boisserie; J. Le Louer; N. Azzabou

G.P.122 **T2-spectrum: A novel NMR approach for the characterization of muscle disorders**
E.C.A. Araujo; P.G. Carlier

G.P.123 **NMR imaging of short T2-components in skeletal muscle tissue**
A. Vignaud; G. Guillot; E.C.A. Araujo; P.G. Carlier

G.P.124 **Increase in T2 relaxation times in skeletal muscle of Duchenne but not Becker muscular dystrophy patients**
B.H. Wokke; J.C. van den Bergen; M.H. Hooijmans; J.J. Verschuuren; E.H. Niks; H.E. Kan

G.P.125 **Manganese enhanced muscle MRI as a Sensitive Outcome Measure of Dystrophin Restoration in the mdx Mouse**
E. Greally; A.M. Blain; S.T. Ahmed; S.H. Laval; A.M. Blamire; V. Straub

G.P.126 **MRI detects Dp140 dystrophin isoform dependent brain changes in boys with DMD**
N. Doorenweerd; C.S.M. Straathof; E.M. Dumas; P. Spitali; H.B. Ginjaar; B.H. Wokke; D.G.M. Schrans; J.C. van den Bergen; E.W. van Zwet; A. Webb; M.A. van Buchem; J.J.G. Verschuuren; J.G.M. Hendriksen; E.H. Niks; H.E. Kan

G.P.127 **Reduced cerebral blood flow in boys with Duchenne muscular dystrophy**
N. Doorenweerd; E.M. Dumas; E. Ghariq; S. Schmid; C.S.M. Straathof; P. Spitali; H.B. Ginjaar; B.H. Wokke; D.G.M. Schrans; J.C. van den Bergen; E.W. van Zwet; A. Webb; M.A.van Buchem; M.J.P. van Osch; J.J.G. Verschuuren; J.G.M. Hendriksen; E.H. Niks; H.E. Kan

G.P.128 **Upper limb muscle MRI fat–water quantification and clinical functional correlation in non-ambulant Duchenne muscular dystrophy**
V. Ricotti; R.B. Evans; C.D.J. Sinclair; J.M. Morrow; J.W. Butler; R.L. Janiczek; M.G. Hanna; P.M. Matthews; T.A. Yousry; F. Muntoni; J.S. Thornton

G.P.129 **Cine-MRI as a new tool to evaluate diaphragmatic dysfunction in Pompe disease**
S.C. Wens; P. Ciet; A. Perez-Rovira; K. Logie; E. Salamon; P. Wielopolski; M. Bruijne; M.E. Kruijshaar; H.W. Tiddens; N.A.M. van der Beek; P.A. van Doorn; A.T. van der Ploeg

G.P.130 **Tracking the brain in myotonic dystrophy: A 5-year longitudinal neuroimaging and neuropsychological follow-up study**
C. Merkel; M. Minnerop; S. Roeske; H. Gaertner; J.C. Schoene-Bake; S. Adler; J.A. Witt; C. Anspach; C. Schneider-Gold; R.C. Betz; C. Helmstaedter; M. Tittgemeyer; K. Amunts; T. Klockgether; B. Weber; C. Kornblum

DMI + Myasthenia + Channel diseases; (G.P.131–141)

Facilitators: Piraye Serdaroglu and Bruno Eymard

G.P.131 **Cell Membrane Integrity in Myotonic Dystrophy Type 1: Implications for Therapy**
A. González-Barriga; J. Kranzen; H.J.E. Croes; W.J.A. van den Broek; B.G.M. van Engelen; J.C.T. van Deutekom; B. Wieringa; S.A.M. Mulders; D.G. Wansink

G.P.132 **Pre-clinical development of peptide-conjugated antisense oligonucleotides for myotonic dystrophy type 1 (DM1)**
S.A.M. Mulders; B. Aguilera; A. Gonzalez-Barriga; J. van de Giessen; W.J.A. van den Broek; D.G. Wansink; J.C.T. van Deutekom; N.A. Datson

G.P.133 **Ankle muscle weakness but not balance trouble account for walking disability in patients with myotonic dystrophy type 1**
G. Ollivier; V. Decostre; I. Ledoux; L. Servais; T. Gidaro; A. Behin; T. Stojkovic; B. Eymard; G. Bassez; J.Y. Hogrel

G.P.134 **Liver functional impairment and glycolipid metabolic abnormality in myotonic dystrophy type 1**
H. Takada; S. Kon; Y. Oyama; T. Kimura; F. Nagahata

G.P.135 **Dental and orthodontic aspects of myotonic dystrophy type 1**
D. Nadaj; A. Lusakowska; D. Maciejak; A.M. Kaminska; M. Zadurska

G.P.136 **Muscle channelopathies: Clinical and genetic features in a large cohort of Italian patients**
L. Maggi; R. Brugnoni; L. Colleoni; D. Kapetis; A. Ardissone; A. Pini; G. Ricci; L. Vercelli; S. Ravaglia; I. Moroni; E. Pegoraro; M. Lo Monaco; V. Sansone; G. Meola; G. Siciliano; T. Mongini; M. Filosto; L. Morandi; R. Mantegazza; P. Bernasconi

G.P.137 **Functional study of five new CLC-1 mutations causing myotonia congenita in Italian families**
P. Imbrici; J.F. Desaphy; R. Brugnoni; L. Colleoni; E. Canioni; D. Kapetis; C. Altamura; P. Bernasconi; L. Morandi; L. Maggi; R. Mantegazza; D. Conte

G.P.138 **Late-onset non-thymomatous generalized myasthenia gravis**
S. Yildiz-Celik; H. Durmus; M. Hajibehzad; V. Yilmaz; P. Oflazer-Serdaroglu; Y. Parman; G. Saruhan-Direskeneli; F. Deyemeer

G.P.139 **Congenital myasthenic syndrome due to novel choline acetyltransferase (ChAT) gene mutations in Kadazandusun family from Borneo**
J.S. Tan; T. Ambang; A. Ahmad-Annuar; K.T. Wong; K.J. Goh

G.P.140 **A novel missense mutation in the AGRN gene causing congenital myasthenic syndrome mimicking neck myopathy**
M. Karakaya; O. Ceyhan-Birsoy; A.H. Beggs; H. Topaloglu

G.P.141 **Dpagt1 mutation: Limb–Girdle congenital myasthenic syndrome due to glycosylation defect**
İ. Öncel; A. Töpf; T. Evangelista; B. Konuşkan; B. Talim; A. Abicht; H. Lochmüller; H. Topaloglu

EDMD + Lamin A/C + Emerin + FHL1; (G.P.142–151)

Facilitators: Susana Quijano-Roy and Joachim Schessl

G.P.142 **A new EMD gene missense mutation in exon 1 leads to absence of emerin and is responsible for X-linked dilated cardiomyopathy with conduction defects and arrhythmias and almost elusive skeletal muscle features**
R. Ben Yaou; M. Gerard; K. Chami; A. Sehier; A. Belin; F. Labombarda; P. Richard; G. Bonne; F. Leturcq; F. Chapon

G.P.143 **Muscle MRI and CT help to differentiate between mutations in emerin and lamin A/C gene in patients with Emery-Dreifuss clinical phenotypes**
J. Díaz-Manera; A. Alejaldre; L. Gonzalez; R. Rojas-García; M. Olivé; J. Llauger; E. Gallardo; L. Gonzalez-Quereda; P. Carbonell; C. Marquez; N. Muelas; J.J. Vílchez; R. Fernández-Torrón; A. Lopez de Munain; I. Illa

G.P.144 **Identification of both LMNA and SMCHD1 mutations in a case with overlapping phenotypes**
T. Stojkovic; P. Richard; P. Charron; S. Rondeau; M. JeanPierre

G.P.145 **LMNA-related muscular dystrophies: Clinical and histopathological spectrum in Argentina**
S. Monges; F. Lubieniecki; F. de Castro; V. Lafuente; M. Gonzalez; G. Reyes; L. Chertkoff; S. Quijano-Roy; N.B. Romero; P. Richard; G. Bonne; P. Guicheney; M. Sacolitti; A.L. Taratuto

G.P.146 **Cardiac Manifestations and Gastro-Intestinal Sequelae in Children with LMNA-CMD**
I. Dabaj; F. Heller; S. Quijano-Roy; J.K. Mah; A. Rutkowski; B. Estournet; R. Clegg; K. Wahbi

G.P.147 **Two novel epsilon isoforms of nesprin-2, a protein linked to Emery-Dreifuss muscular dystrophy**
I. Holt; T.D. Nguyen; L.T. Le; Q. Zhang; C.A. Sewry; C.M. Shanahan; G.E. Morris

G.P.148 **FHL1 mutations are causing familial aortic and other arterial aneurysms with scapuloperoneal myopathy**
J. Schessl; W. Kress; S. Feldkirchner; B. Schoser

G.P.149 **Unique brick-red auto-fluorescence of reducing bodies and protein aggregates is a useful diagnostic biopsy marker for FHL1-associated myopathies**
R. Phadke

G.P.150 **Clinical heterogeneity in adult forms of FHL1 related myopathies. The "Institut de Myologie" experience**
R.A.B. Ben Yaou; T.A.N. Stojkovic; P.A.S. Laforet; A.L.I. De Becdelievre; H.E.N. Becane; K.A.R. Wahbi; C.A.R. Navarro; M.I.C. Fardeau; N.O.R. Romero; P.A.S. Richard; D.E.N. Duboc; G.I.S. Bonne; B.R.U. Eymard

G.P.151 **Loss of *FHL1* function impairs motility and causes myopathy in vivo**
M. Keßler; A. Kieltsch; E. Kayvanpour; B. Schoser; J. Schessl; W. Rottbauer; S. Just

Congenital mypathies; (G.P.152–160)

Facilitators: Kristen Nowak and Edoardo Malfatti

G.P.152 **Novel recessive mutations in *MYH2* presenting with congenital facial weakness, ophthalmoparesis, severe progressive scoliosis, and mild muscle weakness**
M.E. Leach; S. Donkervoort; K. Simpson; A.C. Tesi-Rocha; R. Avery; J. Dastgir; C. Reyes; Y. Hu; T.L. Winder; C.G. Bönnemann

G.P.153 **Recessive myosin myopathy with external ophthalmoplegia associated with MYH2 mutations**
H. Tajsharghi; S. Hammans; C. Lindberg; A. Lossos; N.F. Clarke; I. Mazanti; L.B. Waddell; Y. Fellig; N. Foulds; H. Katifi; O. Raheem; B. Udd; Z. Argov; A. Oldfors

G.P.154 **Mutations in ECEL1 lead to distal arthrogryposis type 5D**
T.G. Whyte; S. Cirak; I. Oprea; P. Beales; D. Osborn U. UK10K consortium2; K. Busch; M. Hurles; C. Longman; R. Quinlivan; C. Sewry; H. Jungbluth; F. Muntoni

G.P.155 **A first Asian MEGF10 myopathy due to novel homozygous mutation**
K. Takayama; S. Mitsuhashi; S. Noguchi; Y.K. Hayashi; I. Nonaka; I. Nishino

G.P.156 **Mutations in FAM111B cause Hereditary Fibrosing Poikiloderma with tendon contracture, myopathy and pulmonary fibrosis**
S. Mercier; S. Küry; A. Magot; N. Bodak; C. Bou-Hanna; V. Cormier-Daire; A. David; L. Faivre; D. Figarella-Branger; R. Gherardi; A. Goldenberg; A. Hamel; J. Igual; D. Israël-Biet; C. Kannengiesser; C. Laboisse; C. Le Caignec; A. Munnich; J.M. Mussini; J. Piard; E. Puzenat; E. Salort-Campana; N. Soufir; C. Thauvin; Y. Péréon; B. Mayosi; S. Barbarot; S. Bézieau

G.P.157 **Clinical and pathological features associated with mutations in MICU1**
A.M. Childs; K. Pysden; H. Roper G. Chow; E.H. Niks; M. Kriek; P.F. Chinnery; D. Lewis-Smith; M. Duchen; G. Szabadkai; C. Logan; E. Sheridan; C. Sewry; F. Muntoni

G.P.158 **A case of centronuclear myopathy, dysmorphisms, short stature, prominent cerebellar folia with mixed central and peripheral signs; can a multisystem condition with autophagy provide a unified explanation ?**
A. Majumdar; A. Fadiliah; K. Vijayakumar; M. Greenslade; K. Kurian

G.P.159 **Cylindrical spirals originate from Sarcoplasmic reticulum of muscles in two Chinese siblings**
C.Z. Yan; J.W. Xu; Y.Y. Zhao; W. Li

G.P.160 **Cysteine mutations cause defective tyrosine phosphorylation in MEGF10 myopathy**
S. Mitsuhashi; H. Mitsuhashi; M.S. Alexander; H. Sugimoto; P.B. Kang

18:00–20:00 **Sarepta Industry Symposium** - Langenbeck-Virchow-Haus (Foyer & Lecture Hall)

Thursday 9 October 2014

08:30–10:00 **Limb-Girdle Muscular Dystrophies (LGMDs);** Invited lectures (L.I.1–3) – Langenbeck-Virchow-Haus
Chairpersons: Katie Bushby and Peter Van den Bergh

L.I.1 **Autosomal dominant limb-girdle muscular dystrophies**
B. Udd

L.I.2 **The ABC of autosomal recessive limb girdle muscular dystrophy**
V. Straub

L.I.3 **Novel disease mechanisms in myopathies**
F. Muntoni

10:00–10:30 **Morning Tea and Coffee** - Langenbeck-Virchow-Haus (Foyer)

10:30–11:00 **Limb-Girdle Muscular Dystrophies (LGMDs);** Invited lecture (L.I.4) – Langenbeck-Virchow-Haus
Chairpersons: Carsten Bönnemann and Marianne de Visser

L.I.4 **Developing therapies in the LGMDs**
K.M. Flanigan

11:00–13:00 **Muscular dystrophies; Oral Presentations (G.O.7–14) –** Langenbeck-Virchow-Haus
Chairpersons: Carsten Bönnemann and Marianne de Visser

G.O.7 **Multiple genetic variations in limb-girdle muscular dystrophies**
M. Savarese; G. Di Fruscio; A. Torella; M. Mutarelli; G.P. Comi; T. Mongini; E. Ricci; C. Angelini; M. Fanin; E. Pegoraro; O. Musumeci; A. Toscano; G. Siciliano; M. Mora; L. Morandi; E.M. Bertini; A. D'Amico; G. Tasca; C. Bruno; C. Fiorillo; C. Minetti; F.M. Santorelli; A. Garofalo; T. Giugliano; C. Pisano; F. Del Vecchio Blanco; G. Piluso; O. De Concilio; S. Sacconi; L. Politano; V. Nigro

G.O.8 **A defect in the RNA-processing protein HNRPDL causes limb-girdle muscular dystrophy 1G (LGMD1G)**
N.M. Vieira; M.S. Naslavsky; L. Licinio; F. Kok; D. Schlesinger; M. Vainzof; N. Sanchez; J.P. Kitajima; L. Gal; N. Cavaçana; P.R. Serafini; S. Chuartzman; C. Vasquez; A. Mimbacas; V. Nigro; R.C.M. Pavanello; M. Schuldiner; L.M. Kunkel; M. Zatz

G.O.9 **AAV gene transfer utilizing homologous overlap vectors mediates functional recovery of dysferlin deficiency**
P.C. Sondergaard; D.A. Griffin; E.R. Pozsgai; R.W. Johnson; J.R. Mendell; L.R. Rodino-Klapac

G.O.10 **Skeletal muscle, cardiac, and pulmonary imaging biomarkers of disease activity in boys with Duchenne muscular dystrophy**
A. Mankodi; R. Janiczek; M. Froeling; N. Azzabou; L. Gaur; D. Stock; R. Evers; C. Bishop; L. Yao; C. Grunseich; A. Arai; P. Carlier; K. Fischbeck

G.O.11 **Longitudinal quantitative muscle MRI in 5 Duchenne boys treated with exon 51 skipping – a pilot study**
M.T. Hooijmans; B.H.A. Wokke; N. Goemans; G. Campion; J.J.G. Verschuuren; E.H. Niks; H.E. Kan

G.O.12 **Semi-automated analysis of diaphragmatic motion during deep breathing using dynamic MRI in both healthy controls and non-ambulant Duchenne muscular dystrophy**
C.A. Bishop; V. Ricotti; C.D.J. Sinclair; J. Butler; R.B.M. Evans; J.M. Morrow; M.G. Hanna; P.M. Matthews; T.A. Yousry; J.S. Thornton; F. Muntoni; R.L. Janiczek

G.O.13 **One year follow-up of Duchenne Muscle Dystrophy with Nuclear Magnetic Resonance imaging and spectroscopy indices**
C. Wary; N. Azzabou; K. Zehrouni; J. Le Louer; M. Montus; T. Voit; L. Servais; P.G. Carlier

G.O.14 **The burden of Duchenne muscular dystrophy: An international, cross-sectional study**
E. Landfeldt; P. Lindgren; C.F. Bell; C. Schmitt; M. Guglieri; V. Straub; H. Lochmuller; K. Bushby

13:00–14:30 **Lunch** – Langenbeck-Virchow-Haus (Foyer)

Meeting of Editorial Board of Neuromuscular Disorders – Rudolf Virchow Saal, Langenbeck-Virchow-Haus (Lunch served)

14:30–16:00 **Guided poster discussion session 3: Parallel sessions (G.P.161–243)** –Thaersaal/ Humboldt Graduate School

DMD 3 – Clinical; (G.P.161–181)
Facilitators: Arpad von Moers and Ros Quinlivan

G.P.161 **Improving the diagnosis of Duchenne muscular dystrophy**
M. Guglieri; H.J.A. van Ruiten; V. Straub; K. Bushby

G.P.162 **The Impact of Reduced Ankle Range of Movement on the Functional Abilities of Patients with Duchenne Muscular Dystrophy**
J.W. Butler; M. Main; F. Muntoni

G.P.163 **Clinical features of Duchenne muscular dystrophy aged over 40 years**
T. Saito; M. Kawai; T. Matsumura; H. Fujimura; S. Sakoda

G.P.164 **Physical ability and health in a non-steroid population of 77 adult patients with Duchenne muscular dystrophy**
B. Werge; J. Rahbek; A. Madsen; J. Marquardt; U. Werlauff; B.F. Steffensen

G.P.165 **Becoming an Adult with Duchenne muscular dystrophy in Canada**
L.C. McAdam; J. Mah; W.D. Biggar

G.P.166 **Comparative economic impact of therapeutic innovation on health care burden of Duchenne Muscular Dystrophy (DMD) using Becker Muscular Dystrophy (BMD) as a comparator for potential clinical outcome corridor**
O. Schreiber; C. Klug; S. Thiele; E. Schorling; J. Zowe; P. Reilich; K. Nagels; M.C. Walter

G.P.167 **Clinical Trial Readiness for Non-Ambulatory Boys and Men with Duchenne Muscular Dystrophy: 12 and 24 Month Follow-up from the MDA-DMD Network**
A.M. Connolly; E.C. Malkus; J.R. Schierbecker; C.A. Siener; P. Anand; J.R. Mendell; K.M. Flanigan; P.T. Golumbek; C.M. Zaidman; C.M. McDonald; E. Henricson; L. Johnson; A. Nicorici;P.I. Karachunski; J.W. Day; J.M. Kelecic; L.P. Lowes; L.N. Alfano; B.T. Darras; P.B. Kang;J.M. Florence

G.P.168 **Time-dependent development of fibrosis and inflammation in Duchenne Muscular Dystrophy**
C. Preuße; U. Schara; H.H. Goebel; N. Zerbe; P. Hufnagl; F.L. Heppner; A. von Moers; W. Stenzel

G.P.169 **Non-fatal fat embolism syndrome in Duchene muscular dystrophy**
L.C. McAdam; K. MacLeod; N. Serrao; W.D. Biggar

G.P.170 **Bone mineral density and body composition in 39 Duchenne muscular dystrophy patients: A two-years follow up**
M.B. Pasanisi; S. Vai; G. Baranello; L. Maggi; I. Moroni; M.T. Arnoldi; C. Bussolino; G. Brenna; M.L. Bianchi; L. Morandi

G.P.171 **Age-specific prevalence of osteoporosis and frequency of poor bone health indices in Duchenne Muscular Dystrophy**
C. Tian; B. Wong; L. Hornung; J. Khoury; L. Miller; J. Bange; I. Rybalsky; M. Rutter

G.P.172 **Renal function in children and adolescents with Duchenne muscular dystrophy: A prospective study**
L. De Waele; E. Braat; P. Vermeersch; O. Gheysens; E. Levtchenko; H. Pottel; L. Hoste; N. Goemans

G.P.173 **Are current formulas for estimated glomerular filtration rate reliable in children and adolescents with Duchenne muscular dystrophy?**
L. De Waele; E. Braat; P. Vermeersch; O. Gheysens; E. Levtchenko; H. Pottel; L. Hoste; N. Goemans

G.P.174 **Behavioral and neurocognitive profile in Duchenne Muscular Dystrophy**
P. Colombo; F. Civati; E. Mani; S. Gandossini; E. Brighina; G.P. Comi; N. Bresolin; A.C. Turconi; M. Molteni; M. Nobile; M.G. D'Angelo

G.P.175 **Cognitive and neurobehavioral profile and its relation with genotype mutation in boys with Duchenne muscular dystrophy**
R. Banihani; S. Smile; G. Yoon; M. Mosleh; A. Snider; L.C. McAdam

G.P.176 **Steroid therapy and respiratory function in Duchenne muscular dystrophy**
M.G. D'Angelo; A. LoMauro; M. Romei; S. Gandossini; E. Brighina; E. Marchi; N. Bresolin; A. Aliverti

G.P.177 **Constipation in Duchenne muscular dystrophy: Common, underdiagnosed and undertreated**
D. Kraus; B.L. Wong; S.Y. Hu; P. Horn; I. Rybalsky; K.C. Shellenbarger; A. Kaul

G.P.178 **BMI but not height correlates with timed motor function tests of DMD patients on long term daily glucocorticoid therapy**
B.L. Wong; H.N. Lee; P. Horn; S.Y. Hu; J. Bange; B. Godshall; I. Rybalsky

G.P.179 **Association between resting energy expenditure and body weight change in patients with Duchenne muscular dystrophy**
S. Baba; S. Takanoha; A. Ishiyama; H. Komaki; E. Takeshita; H. Imaizumi; Y. Abe; M. Kobayashi; Y. Kumazawa; M. Sasaki

G.P.180 **Evaluation of cardiologic status in Carriers of Duchenne Muscular Dystrophy**
A. Schoenecker; J. Schelhorn; T.W. Schlosser; U. Neudorf; U. Schara

G.P.181 **Early onset and severe X-linked dilated cardiomyopathy with epilepsy, behavioral abnormalities and elevated CK due to DMD gene intron 1 splice site mutation**
R. Ben Yaou; J.U.L. Nectoux; F.R.A. Iserin; S.A.L. Ould Amar; N.O.R. Romero; S.H.A. Varnous; F.R.A. Leturcq; P.A.S. Laforet

16.00–17.30 **Outcome Measures 1;** (T.P.1–14)

Facilitators: Laurent Servais and Nathalie Goemans

T.P.1 **Pilot study evaluating motivation on the performance of timed walking in boys with Duchenne muscular dystrophy**
L.N. Alfano; L.P. Lowes; K.M. Berry; H. Yin; I. Dvorchik; K.M. Flanigan; L. Cripe; J.R. Mendell

T.P.2 **The age of ambulation in boys with Duchenne muscular dystrophy and its use as an end-point in clinical trials**
J.J. Giss; T. Johnson; D.J. Fox; A. Kumar; E. Ciafaloni; S. Kim; M. Yang; A.J. van Essen; R.S. Finkel

T.P.3 **Gait velocity in boys with Duchenne muscular dystrophy -impact of overweight, muscle strength and range of motion on gait velocity**
L. Wahlgren; A.K. Kroksmark

T.P.4 **Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36 month changes**
E.S. Mazzone; M. Pane; S. Sivo; C. Palermo; M.P. Sormani; S. Messina; A. D'Amico; G.L. Vita; L. Fanelli; A. Berardinelli; M.A. Donati; G. Baranello; R. Battini; E. Pegoraro; L. Politano; C. Bruno; G.P. Comi; E. Bertini; E. Mercuri

T.P.5 **Timed function tests and other physical function outcomes in Ataluren-treated patients with nonsense mutation Duchenne Muscular Dystrophy (nmDMD)**
C. McDonald; A. Reha; G.L. Elfring; S.W. Peltz; R. Spiegel

T.P.6 **Six minute walk test: Reference values and prediction equation in healthy boys aged 5 to 12 years**
N. Goemans; K. Klingels; M. vanden Hauwe; H. Feys; G. Buyse

T.P.7 **Prediction equation to estimate 6-Minute-Walk-Distance from motor function tests in patients with Duchenne Muscular Dystrophy**
B.L. Wong; S.Y. Hu; P. Horn; P. Morehart; M. McGuire; A. McCormick

T.P.8 **PODCI subscales are sensitive to differences in disease stage and disease progression in DMD**
E.K. Henricson; A. Cnaan; R.T. Abresch; C.M. McDonald; A. The CINRG Investigators

T.P.9 **Development of a cloud computing for Holter movement analysis in neuromuscular diseases**
A.G. Le Moing; A. Totoescu-Seferian; A. Moraux; E. Dorvaux; N. Porteix; M. Annoussamy; C. Pelegry; A. Taibi; D. Vissière; J.Y. Hogrel; L. Servais

T.P.10 **6 minute walk test and Performance of Upper Limb in ambulant DMD boys**
M. Pane; C. Palermo; S. Sivo; E. Mazzone; L. Fanelli; R. De Sanctis; A. D'Amico; S. Messina; L. Politano; R. Battini; M. Pedemonte; E. Pegoraro; G. D'Angelo; A. Pini; G. Baranello; E. Mercuri

T.P.11 **Longitudinal assessment of Upper Limb function in DMD patients: 12 month changes**
M. Pane; E.S. Mazzone; L. Fanelli; R. De Sanctis; C. Palermo; S. Sivo; A. D'Amico; S. Messina; L. Politano; E. Battini; M. Pedemonte; E. Pegoraro; A.L. Berardinelli; G. D'Angelo; A. Pini; G. Baranello; E. Mercuri

T.P.12 **The Performance of Upper Limb scores correlate with pulmonary function test measures and Egen Klassifikation scores in Duchenne muscular dystrophy**
H.N. Lee; H. Sawnani; P. Horn; L. Relucio; B.L. Wong

T.P.13 **Upper limb performance changes during a one-year follow-up in non-ambulant patients with Duchenne muscular dystrophy**
A.M. Seferian; A. Moraux; M. Annoussamy; A. Canal; V. Decostre; O. Diebate; A.G. Le Moing; T. Gidaro; N. Deconinck; F. Van Parys; W. Vereecke; S. Wittevrongel; M. Mayer; K. Maincent; I. Desguerre; C. Themar-Noel; J.M. Cuisset; V. Tiffereau; S. Denis; V. Jousten; S. Quijano-Roy; T. Voit; J.Y. Hogrel; L. Servais

T.P.14 **Arm elevation assessment (area) for Duchenne muscular dystrophy: Preliminary study of a practical instrument for professionals**
A.A. Karaduman; I. Alemdaroglu; O. Yilmaz; H. Topaloglu

Mitochondrial Disease; (G.P.182–194)

Facilitators: Mar Tulinius and Ekkehard Wilichowski

G.P.182 **Severe and progressive mitochondrial myopathy in early childhood: Novel mutations in TK2 gene**
A. Nascimento; J. Dominguez; R. Mati; C. Ortez; M. Madruga; S. Emperador; J. Montoya; J. Aguirre; C. Jou; J. Colomer; C. Jimenez- Mallebrera

G.P.183 **TK2 mutation: An expanding clinical phenotype**
D. Ram; M.I. Hughes

G.P.184 **Mitochondrial DNA depletion in single fibers in a patient with novel *TK2* mutations**
S. Roos; U. Lindgren; C. Ehrstedt; A.R. Moslemi; A. Oldfors

G.P.185 **The novel non-sense mutation m.4214G > A in MT-ND1 results in mitochondrial myopathy with severe complex I deficiency**
D. Ronchi; M. Sciacco; A. Bordoni; I. Colombo; D. Piga; F. Fortunato; M. Moggio; G.P. Comi

G.P.186 **Gene expression profile of cybrid cells harbouring a mitochondrial DNA mutation in the MT-ATP6 gene reveals new pathogenic pathway**
G. Fayet; K. Aure; P. Lesimple; C. L'Hermitte-Stead; C. Chevalier; A. Magot; R. Houlgatte; Y. Pereon; A. Lombes; F. Savagner

G.P.187 **MTO1 mutation in type 2 fiber lipid storage myopathy**
M.K. Bakhshandeh Bali; S. Noguchi; I. Nishino

G.P.188 **Autosomal recessive Kearns-Sayre syndrome in a girl with altered mitochondrial DNA transcription caused by RRM2B gene defect**
E.K.G. Wilichowski; A. Abicht; H. Mayr; R. Horvath; W. Sperl; J. Gärtner

G.P.189 **Exercise intolerance associated with atypical facial muscle hypertrophy related to mitochondrial tRNA(Pro)gene mutation**
Y. Péréon; A. Magot; G. Fayet; S. Mercier; J.M. Mussini; K. Auré; A. Lombès; C. Jardel

G.P.190 **Multiple deletions in mitochondrial DNA in myofibrillar myopathy and centronuclear myopathy**
J. Schaefer; U. Reuner; H. Reichmann; M. Meinhardt; S. Jackson

G.P.191 **What factors are associated with the prevalence of sub-sarcolemmal mitochondrial aggregates (SSMA) in paediatric skeletal muscle? Examining the use and limitations of SSMA as a diagnostic muscle biopsy marker**
A. Cortese; M. Ellis; C. Fratter; Z. Fox; D. Chambers; P. Hodsdon; I. Hargreaves; M. Kinali; S. Rahman; C. Sewry; F. Muntoni; J. Poulton; R. Phadke

G.P.192 **Muscle heart brain disease in 3 of 4 siblings**
J. Radke; K. von Au; M. Dreesmann; H. von Pein; W. Stenzel; H.H. Goebel

G.P.193 **Ataxia, core myopathy, beta-ketothiolase deficiency, dentate nuclear abnormalities and learning difficulties in a pair of consanguineous siblings. A new association or double trouble?**
A. Majumdar; G. Pierre; A. Bowron; S. Love; K. Kurian

G.P.194 **Intramuscular variation in mitochondrial functionality and sarcoplasmic proteome profile of bovine semimembranosus muscle**
M.N. Nair; S.P. Suman; R. Ramanathan; M.K. Chatli; S. Li; P. Joseph; C.M. Beach; G. Rentfrow

Metabolic myopathies; (T.P.15–34)

Facilitators: Antonio Toscano and Ans van der Ploeg

T.P.15 **Enzymotherapy in late onset Pompe disease patients: A 4-year longitudinal study using quantitative MRI**
E. Doche; E. Salort-Campana; A. Le Troter; S. Attarian; J. Pouget; D. Bendahan

T.P.16 **Clinical and molecular characteristics of 33 patients with Late Onset Pompe Disease (LOPD): Unusual phenotypes, novel mutations and therapeutic responses**
F. Montagnese; O. Musumeci; E. Barca; S. Romeo; A. Ciranni; M. Aguennouz; C. Rodolico; A. Toscano

T.P.17 **Asymptomatic Pompe disease: A study of 6 patients**
P. Laforêt; R.Y. Carlier; K. Laloui; P. Carlier; E. Salort-Campana; J. Pouget; A. Echaniz-Laguna

T.P.18 **Late-onset Pompe Disease: Histopathological, biochemical and clinical assessment before and after ERT**
M. Sciacco; D. Ronchi; M. Ripolone; R. Violano; V. Lucchini; R. Xhani; G.P. Comi; F. Fortunato; A. Bordoni; P. Tonin; M. Filosto; S. Previtali; T. Mongini; L. Vercelli; E. Vittonatto; A. Toscano; O. Musumeci; E. Barca; C. Angelini; C. Lamperti; M. Mora; L. Morandi; M. Moggio

T.P.19 **Effects of antibody formation during enzyme replacement therapy in 73 adult patients with Pompe disease**
J.M. de Vries; E. Kuperus; M. Hoogeveen-Westerveld; S.C.A. Wens; M.A. Kroos; M.E. Kruijshaar; P.A. van Doorn; A.T. van der Ploeg; W.W.M. Pijnappel

T.P.20 **Effect of Enzyme replacement therapy in Late onset Pompe Disease: Open pilot study of 60 weeks follow up**
J.S. Park; Y.E. Park; Y.C. Choi; J.H. Shin; J.M. Lee; D.S. Kim

T.P.21 **Assessing immunogenicity of rhGAA in adult Pompe disease subjects**
E. Masat; P. Laforet; D. Amelin; P. Veron; K. Laloui; O. Benveniste; F. Mingozzi

T.P.22 **Two cases of late-onset Pompe disease treated with enzyme replacement therapy: Clinical outcome for 3 years follow-up**
J.H. Lee; Y.N. Cho; H.J. Park; Y.C. Choi

T.P.23 **Search for Pompe patients in patients with undetermined myopathies in South-West Sweden**
C. Lindberg; B. Andersson; A. Oldfors

T.P.24 **Alglucosidase Alfa Reduces Lysosomal Glycogen in Skeletal Muscle Biopsies of Patients with Late-Onset Pompe Disease (LOPD)**
B.L. Thurberg; A. van der Ploeg; J.T. Kissel; B. Schoser; A. Pestronk; R.J. Barohn; O. Goker-Alpan; T. Mozaffar; L.D.M. Pena; Z. Simmons; V. Straub; P. Young; C. Bjartmar

T.P.25 **The benefit of attending a McArdle disease multidisciplinary clinic: Preliminary results on two outcome measures**
S. Chatfield; J. Pattni; C. Ellerton; R. Carruthers; R. Godfrey; R. Quinlivan

T.P.26 **McArdle disease misdiagnosed as meningitis: A case report**
R.S. Scalco; S. Chatfield; J. Pattni; C. Ellerton; A. Beggs; R. Godfrey; J.L. Holton; R. Quinlivan

T.P.27 **Diagnostic power of the non-ischemic forearm exercise test in glycogenosis type V**
F. van den Bogaart; P. Laforêt; I. Ledoux; F. Petit; N. Koujah; A. Béhin; T. Stojkovic; B. Eymard; N. Voermans; J.Y. Hogrel

T.P.28 **The Significance of Clinical and Laboratory features in the diagnosis of Glycogen Storage Disease Type V: A Case Report**
H.J. Park; J.H. Lee; B.C. Suh; H.Y. Shin; Y.C. Choi

T.P.29 **Role of AMP deaminase and adenylate kinase in production of ammonia in skeletal muscle of patients with McArdle's disease**
P.R. Joshi; T. Apitz; S. Zierz

T.P.30 **Recurrent episodes of myoglobinuria, mental retardation and epilepsy but no haemolysis in two brothers with phosphoglycerate kinase deficiency**
S. Coppens; N. Deconinck; R. van Wijk; P. Koralkova; P. Van Bogaert; A. Aeby

T.P.31 **Neurite length can be an outcome parameter for drug screening in multiple acyl-CoA dehydrogenase deficiency**
Y.F. Lin; D.E. Liu; I. Nishino; Y.J. Jong; W.C. Liang

T.P.32 **Sensory neuropathy as a major clinical feature of LCHAD deficiency**
S. Souvannanorath; T. Maisonobe; V. Valayannopoulos; Y. Nadjar; F. Mochel; A. Boutron-Corriat; M. Brivet; P. Laforêt

T.P.33 **Clinical and genetic characterization of patients with repeated rhabdomyolysis**
M. Cabrera; R. Ghaoui; D. Mourdant; P.J. Lamont; N. Clarke; N.G. Laing

T.P.34 **A family with epilepsy, movement disorders, mental retardation and exercise-induced myoglobinuria: A complex phenotype caused by two different rare disorders**
A. Toscano; E. Ferlazzo; S. Romeo; F. Montagnese; U. Aguglia; C. Rodolico; O. Musumeci

Aging and Muscle Homeostasis; (G.P.195–206)

Facilitators: Helge Amthor and Baziel van Engelen

G.P.195 **BMP signaling controls satellite cell dependent postnatal muscle growth**
C. Beley; E. Schirwis; E. Mouisel; S. Alonso-Martin; A. Rochat; L. Garcia; A. Ferry; F. Relaix; M. Schuelke; F. Le Grand; H. Amthor

G.P.196 **Metabolic effects of the Compact myostatin**
T. Kocsis; J.A. Baan; L. Mendler; L. Dux; A. Keller-Pinter

G.P.197 **Strongman syndrome: Clinical, pathological and genetic characterization of dominant herculean myalgic disorders**
T.C. Conte; M. Tetreault; M.J. Dicaire; S.M. Provost; N. Al-Bustani; B. Beland; M.P. Dube; V. Bolduc; M. Srour; E. O'Ferrall; J.P. Bouchard; G. Ravenscroft; N. Laing; P. Lamont; J. Mathieu; R.T. Hepple; B. Brais

G.P.198 **Selenoprotein N and oxidative stress are novel regulators of myogenesis and muscle cell stemness**
S. Arbogast; J. Rowell; A. Pannérec; C. Serreri; C. Ramahefasolo; A. Ferreiro

G.P.199 **In vitro engineered muscle: Identification and characterization of murine dermal precursor cells with myogenic potential**
P. García-Parra; N. Naldaiz-Gastesi; M. Goicoechea; S. Alonso-Martín; A. Aiastui; M. López-Mayorga; P. García-Belda; J. Lacalle; V. Le Berre; A. Matheu; J.M. García-Verdugo; J.J. Carvajal; F. Relaix; A. López de Munain; A. Izeta

G.P.200 **The expression profile of developmental markers in diagnostic muscle biopsies indicates the presence of a "neurogenic muscle regeneration"**
O. Danielsson; L. Gröntoft; B. Häggqvist; I. Gati; M. Vrethem; J. Ernerudh

G.P.201 **Aberrant mechanical-metabolic coupling in muscular dystrophy: Gene expression and functional studies in mdx mouse muscle in relation to age and exercise**
G.M. Camerino; R.F. Capogrosso; M. Cannone; P. Mantuano; A. Giustino; A.M. Massari; A. Cozzoli; R.W. Grange; A. De Luca

G.P.202 **Characterization of skeletal muscle and heart during aging in rats, and response to formoterol, a selective β2 adrenoceptor agonist, in aged rats**
C. Lambert; J. Boesch; M. Obrecht; J. Grosjean; P.R. Allegrini; S. Hatakeyama

G.P.203 **IL-6- and calcineurin-mediated but not IGF-1-mediated mechanisms contribute to the upregulation of MHC I and HSP70 mRNA levels in C2C12 cells**
Y. Mori; J. Yamaji; R. Hiroshima; T. Nakano; A. Miyazaki; M. Watanabe

G.P.204 **The effect of water-soluble fullerene in muscle regeneration process**
A. Ishii; M. Yoshida; N. Ohkoshi; H. Ueno; K. Kokubo; A. Tamaoka

G.P.205 **A Modified Cysteine Knot Ligand Trap of the TGFβ Superfamily, ACE-083, Increases Muscle Mass Locally in a Mouse Model of Duchenne Muscular Dystrophy**
A. Mulivor; D. Sako; M. Cannell; S. Wallner; K. Hevron; R. Steeves; R. Castonguay; R.S. Pearsall; R. Kumar

G.P.206 **Consequences of dietary challenge and aging on macroautophagy in Myf5^{+} cells and muscle**
J. Sarparanta; N. Martinez-Lopez; S. Sahu; R. Singh

CMD and Col VI; (G.P.207-218)

Facilitators: Caroline Sewry and Enrico Bertini

G.P.207 **Clinical and genetic spectrum in a large cohort of patients with a genetic diagnosis of Congenital Muscular Dystrophies: Analysis of the UK diagnostic service 2001-2013**
M. Sframeli; A. Sarkozy; G. Astrea; M. Scoto; L. Feng; R. Mein; M. Yau; R. Phadke; C. Sewry; S. Messina; S. Robb; F. Muntoni

G.P.208 **Whole body MRI in SEPN1-related myopathy shows an homogeneous and recognisable pattern**
I. Dabaj; K. Hankiewicz; R. Carlier; L. Lazaro; J. Linzoain; C. Barnerias; D. Avila-Smirnow; D. Gomez Andres; A. Ferreiro; B. Estournet; P. Richard; S. Bulacio; S. Quijano-Roy

G.P.209 **N-acetylcysteine as an effective treatment *in vivo* and identification of biomarkers in SEPN1-related myopathy: A first preclinical trial**
S. Arbogast; C. Dill; C. Ramahefasolo; F. Piemonte; C. Serreri; A. Lescure; A. Ferry; M. Bonay; E. Bertini; A. Ferreiro

G.P.210 **Gene expression profile of satellite cells from Large^{myd} and Lama2^{dy2j}/J, murine models for muscular dystrophies**
P.C.G. Onofre-Oliveira; P.C.M. Martins; C.F. Almeida; A. Lanzotti; M. Vainzof

G.P.211 **Combination therapy with Glatiramer acetate and FTS improves strength and fibrosis in the dy^{2J}/dy^{2J} mouse model of MDC1A**
N. Yanay; I. Kasis; M. Elbaz; S. Laban; B. Issa; M. Rabie; S. Mitrani-Rosenbaum; Y. Nevo

G.P.212 **Mechanisms and therapeutic approaches to counteract MDC1A: The role of laminin self-assembly and linkage to muscle membrane**
S. Meinen; G. Maier; M. Chauhan; S. Lin; K.K. McKee; S.C. Crosson; P.D. Yurchenco; M.A. Ruegg

G.P.213 **Investigating the molecular mechanisms underlying the progression of collagen VI-related muscular dystrophies**
E. Guadagnin; J. Dastgir; L. Yang; K. Johnson; Q. Wang; Y. Hu; A. Dillmann; M. Cookson; C. Bönnemann

G.P.214 **Mosaicism for dominant COLVI mutations as a cause for intra-familial phenotypic variability**
Y. Hu; S. Donkervoot; T. Stojkovic; N. Voermans; A.R. Foley; M. Leach; J. Dastgir; V. Bolduc; T. Cullup; A. Becdelièvre; L. Yang; H. Su; K. Meilleur; A. Schindler; E. Kamsteeg; P. Richard; R. Butterfield; T. Winder; T. Crawford; R. Weiss; F. Muntoni; V. Allamand; C. Bönnemann

G.P.215 **Ullrich congenital muscular dystrophy: A new cellular study on cultured skin fibroblasts**
N. Deconinck; B. De Paepe; S. Symoens; A. Vanlander; C. Gartioux; V. Allamand; J. Smet; B. Devreese; R. Van Coster

G.P.216 **Allele-specific silencing of a dominant-negative mutation using siRNA or LNA antisense oligonucleotides alleviates the phenotype of a cellular model of Ullrich congenital muscular dystrophy**
V. Bolduc; Y. Zou; M. Lindow; S. Obad; C.G. Bönnemann

G.P.217 **Activation of mesenchymal progenitor cells in skeletal muscles of Collagen VI deficient mice**
S. Noguchi; M. Ogawa; I. Nishino

G.P.218 **Role of Collagen VI as a soluble factor in the control of glucose and adipose tissue homeostasis**
O. Osorio-Conles; M.A. Rodriguez; S. Paco; A. Nascimento; A.M. Gómez-Foix; C. Jimenez-Mallebrera

LGMD 1; (G.P.219–230)

Facilitators: David Hilton-Jones and Mayana Zatz

G.P.219 **Diagnosing the limb-girdle muscular dystrophies using whole exome sequencing: An Australian cohort**
R. Ghaoui; A. Corbett; M. Needham; M. Farrar; H. Sampaio; D. Mowat; S. Rajagopalan; C. Liang; S. Kaur; L. Waddell; K. Daly; B.P. Thomas; M. Lek; M.J. Daly; K.N. North; D.G.MacArthur; C.M. Sue; N.F. Clarke

G.P.220 **Are all the previously reported genetic variants in limb girdle muscular dystrophy genes pathogenic?**
G. Di Fruscio; M. Savarese; A. Garofalo; M. Mutarelli; V. Nigro

G.P.221 **Diagnostic Yield for LGMD at the LHSC Muscle Clinic**
A.G. Florendo-Cumbermack; M. Tripic; S.L. Venance

G.P.222 **Limb girdle muscular dystrophy in the Czech Republic**
K. Stehlikova; D. Skalova; Z. Hruba; L. Fajkusova

G.P.223 **Heart and Limb-Girdle Muscular Dystrophies**
M. Scutifero; A. Taglia; P. D'Ambrosio; R. Petillo; L. Passamano; A. Palladino; V. Nigro; G. Nigro; L. Politano

G.P.224 **A regional panorama of sarcoglycanopathies**
G. Diniz; F. Hazan; H.T. Yildirim; A. Unalp; M. Polat; G. Serdaroglu; S. Ture; G. Akhan; A. Tukun

G.P.225 **Concomittant alpha and gamma sarcoglycan deficiencies in a Turkish boy with a novel deletion in the alpha sarcoglycan gene**
G. Diniz; H.T. Yildirim; S. Gokben; G. Serdaroglu; F. Hazan; K. Yararbas; A. Tukun

G.P.226 **Sarcolemmal deficiency of sarcoglycan complex in an eighteen months old Turkish boy with a huge deletion in the beta sarcoglycan gene**
G. Diniz; H. Tekgul; F. Hazan; K. Yararbas; A. Tukun

G.P.227 **Alpha-Sarcoglycan gene transfer leads to functional improvement in a model of LGMD2E**
E.R. Pozsgai; D.A. Griffin; K.N. Heller; J.R. Mendell; L.R. Rodino-Klapac

G.P.228 **Exon 7 deletion in gamma-sarcoglycan gene: Evidence of a founder effect in Southern Italy**
P. D'Ambrosio; E. Picillo; R. Petillo; A. Taglia; A. Torella; V. Nigro; L. Politano

G.P.229 **The relationship of calpain 3 and titin in the M-band**
K. Charton; J. Sarparanta; A. Vihola; L. Suel; N. Daniele; P. Hackman; B. Udd; I. Richard

G.P.230 **Clinical and genetic features in Korean patients with CAPN3 mutations**
H.J. Park K.D. Park; J.H. Lee; D.S. Shim; Y.C. Choi

SMA; (G.P.231–243)

Facilitators: Helen Roper and Martine Barkats

G.P.231 **A novel splice-site mutation in SMN1 resulting in a very severe SMA1 phenotype**
D. Ronchi; S. Previtali; F. Magri; S. Corti; G.P. Comi

G.P.232 **Double trouble: A child with spinal muscular atrophy type III and Pompe disease**
E. Hobbiebrunken; W. Schulz-Schaeffer; T. Podskarbi; C.R. Mueller-Reible; L. Klinge; H.H. Goebel; E. Wilichowski

G.P.233 **Neuroblastoma in a patient with spinal muscular atrophy (SMA) Type I: Is it just a coincidence?**
E. Sag; H. Susam Sen; G. Haliloglu; B. Yalcin; T. Kutluk

G.P.234 **A family with late-onset spinal muscular atrophy with remarkable phenotypic variability, influenced by SMN2 copy number**
J.S. Park; G.H. Kim; D.S. Kim; J.H. Shin

G.P.235 **Growth and endocrinological evaluation in spinal muscular atrophies – A single centre study of 43 pediatric patients**
H. Trippe; A. Boukidis; B. Hauffa; K. Konrad; U. Schara

G.P.236 **Two siblings with SMARD1, one of them being in "double trouble"**
M. Rasmussen; B. Karime; I.L. Matthews; K. Ørstavik

G.P.237 **Autosomal-dominant spinal muscular atrophy due to mutations in BICD2 gene in Bulgarian patients**
K. Peeters; I. Litvinenko; T. Chamova; B. Asselbergh; L. Almeida-Souza; T. Geuens; E. Ydens; M. Zimon; J. Irobi; E. De Vriendt; V. De Winter; T. Ooms; V. Timmerman; I. Tournev; A. Jordanova

G.P.238 **Phenotypic spectrum of three patients affected by TRPV4 mutations**
C. Ortez; J. Fernandez; E. López Laso; A. Nascimento; M. Olivé; C. Jou; C. Jimenez Mallebrera; F. Baas; J. Colomer

G.P.239 **Expanding phenotype of TRPV4 related neuropathies with notable intrafamilial variability**
L. Medne; C. Bönnemann; S. Scherer; R.S. Finkel; X. Ortiz-Gonzalez; A. Glanzman; T. Estilow; A. Moll; R. Leshner; Y. Wang; T. Winder; S. Yum

G.P.240 **Next Generation Sequencing reveals IGHMBP2 variants as a cause for distal hereditary motor neuronopathy in two children**
A. Majumdar; K. Vijayakumar; A. Merrison; C. Buxton; S. Burton-Jones; C. Crosby; T. Antoniadi

G.P.241 **Muscle histopathology in late-onset spinal muscular atrophies differs from c9orf72-related-ALS**
M.E. Jokela; S. Huovinen; J. Palmio; S. Penttilä; B. Udd

G.P.242 **A trial of hybrid assistive limb (HAL) for a spinal muscular atrophy (SMA) patient**
Y. Iwata; T. Saito; H. Nagayama; H. Yamamoto; H. Nishizono; K. Shibuichi; K. Inoue; H. Fujimura; T. Nakajima

G.P.243 **Orthosis usage and ambulation levels in different clinical types of SMA**
S. Subasi; T.I. Yildiz; N. Bulut; I. Alemdaroglu; A.A. Karaduman; O.T. Yilmaz; H. Topaloglu

16:00–17:30 **Guided poster discussion session 4: Parallel sessions (G.P. 244–321)** – Thaersaal/Humboldt Graduate School
Afternoon Tea and Coffee

Registries and databases; (G.P.244–259)
Facilitators: Jan Verschuuren and Janbernd Kirschner

G.P.244 **The development and implementation of a Managed Clinical Neuromuscular Network in the Southwest of the United Kingdom. The first five years**
A. Majumdar; K. Vijayakumar; A. Merrison; E. Househam

G.P.245 **Exercise related muscle disorders: The EUROMAC Registry for McArdle disease and other rare glycogenolytic disorders**
R.S. Scalco; R. Quinlivan; R. Martin; N. Baruch; M. Martin; C. Navarra; A. Martinuzzi; C. Bruno; P. Laforet; S. Sacconi; A. Wakelin; G. Hadjigeorgiou; J. Vissing; M. Vorgerd; R. Haller; Z. Oflazer; J. Pouget; A. Lucia; T. Andreu

G.P.246 **Prevalence of neuromuscular disorders in the paediatric population in Yorkshire: Variation by ethnicity**
I. Woodcock; K. Pysden; S. Manning; L.K. Taylor; A.M. Childs

G.P.247 **International Clinical Outcome Study in Dysferlinopathy (COS): Results of screening questionnaires in UK patients**
E.A. Harris K. Bettinson; M. James; A. Mayhew; M. Eagle; K. Bushby

G.P.248 **Clinical trial readiness of paediatric neuromuscular patients in Ireland**
A.R. Foley; C. Coman; A. Tobin; N. Kehoe; B. Shinners; A. Timothy; M. McGrath; M. Goode; B. Lynch

G.P.249 **Frequency of multisystem abnormalities among Czech patients with myotonic dystrophy**
O. Parmova; S. Vohanka; J. Strenkova

G.P.250 **Czech National Registries of Hereditary Neuromuscular Disorders**
S. Vohanka; O. Parmova; R. Mazanec; P. Vondracek; L. Mrazova; J. Haberlova; M. Brazdilova; J. Strenkova; P. Brabec

G.P.251 **The Italian Registry of Limb Girdle Muscular Dystrophy: Natural history, genotype-phenotype correlations and outcome measures**
F. Magri; A. Govoni; R. Brusa; C. Angelini; M.G. D'Angelo; T. Mongini; A. Toscano; G. Siciliano; G. Tomelleri; M. Mora; V. Nigro; E. Pegoraro; L. Morandi; O. Musumeci; M. Sciacco; G. Ricci; I. Moroni; S. Gandossini; R. Del Bo; F. Fortunato; D. Ronchi; S. Corti; M. Moggio; N. Bresolin; G.P. Comi

G.P.252 **Beta-sarcoglycanopathy: Any longer an "orphan" disease?**
R. Maggi; B. Vola; M. Cerletti; P. Bonetti

G.P.253 **The Belgian Neuromuscular Disease Registry, results of the 2012 data**
A. Roy P. Van den Bergh; P. Van Damme; V. Van Casteren

G.P.254 **Epidemiology of limb-girdle muscular dystrophies in Sweden**
H. Balcin; C. Lindberg; A. Sundström; G. Solders

G.P.255 **Epidemiology and screening of Pompe disease in Sweden**
H. Balcin; C. Lindberg; A. Sundström; M. Hult; M. Engvall; G. Solders

G.P.256 **The German patient registry for inclusion body myositis**
O. Schreiber; S. Krause; S. Thiele; M. Kiel; M. Vorgerd; J. Schmidt; M.C. Walter

G.P.257 **Prevalence of Neuromuscular Disorders – A systematic review**
A. Theadom; M.J. Rodrigues; S. Ballala; R. Bhattacharjee; C. Higgins; K. Jones; R. Krishnamurthi; V. Feigin

G.P.258 **National registry of Japanese dystrophinopathy patients: Remudy**
F. Takeuchi; H. Nakamura; S. Mitsuhashi; M. Mori-Yoshimura; Y.K. Hayashi; R. Shimizu; H. Komaki; I. Nishino; M. Kawai; S. Takeda; E. Kimura

G.P.259 **Nationwide Patient registry of GNE myopathy in Japan**
M. Mori-Yoshimura; Y.K. Hayashi; N. Yonemoto; M. Murata; S. Takeda; I. Nishino; E. Kimura

Nemaline myopathies; (G.P.260–274)

Facilitators: Carina Wallgren-Petterson and Ana Lia Taratuto

G.P.260 **Clinical diversity in patients with nemaline myopathy**
J.M. Lee; S.J. Hwang; Y.E. Park; J.H. Shin; D.S. Kim

G.P.261 **Whole exome sequencing in patients with congenital myopathies**
I.T. Zaharieva; I. Colombo; M. Sframeli; J.H. Sigurðsson; L. Feng; R. Phadke; C.A. Sewry; J.E. Morgan; F. Muntoni

G.P.262 **Selectivity patterns on lower limb skeletal muscle imaging in patients with nemaline myopathy**
M. Okubo; A. Ishiyama; H. Komaki; E. Takeshita; T. Saito; Y. Saito; E. Nakagawa; K. Sugai; Y.K. Hayashi; I. Nishino; M. Sasaki

G.P.263 **The role of rods in nemaline myopathy and the cause of muscle weakness**
T.E. Sztal; M. Zhao; C. Williams; V. Oorschot; A. Parslow; T.E. Hall; A. Costin; G. Ramm; P.D. Currie; N.G. Laing; K.J. Nowak; R.J. Bryson-Richardson

G.P.264 **Congenital myopathies with protein aggregates and inclusions: Importance of extensive muscle biopsy analysis in the diagnostic workup**
E. Malfatti; S. Monges; J. Bohm; S. Quijano-Roy; G. Brochier; F. Lubieniecki; A. Behin; P. Laforêt; T. Stojkovic; M.T. Viou; M. Beuvin; B. Estournet; B. Eymard; A.L. Taratuto; J. Laporte; M. Fardeau; N.B. Romero

G.P.265 **Nebulin-related nemaline myopathy: Clinical and histopathological spectrum in Argentinean patients**
S. Monges; F. Lubieniecki; F. de Castro; J. Mozzoni; V. Leske; P. Gravina; M. Saccoliti; E. Malfatti; J. Bohm; J. Laporte; N.B. Romero; A.L. Taratuto

G.P.266 **Large copy number variations in *NEB* are frequent in nemaline myopathy patients**
K. Kiiski; V.L. Lehtokari; L. Laari; C. Wallgren-Pettersson; K. Pelin

G.P.267 **Nebulin-associated myopathy: New genetic and pathologic presentations**
Y.E. Park; S.H. Park; J.M. Lee; J.H. Shin; B.R. Kang; C.H. Lee; D.S. Kim

G.P.268 **Whole body muscular MRI in the NEB mutated nemaline myopathy**
I. Dabaj; S. Quijano-Roy; B. Estournet; B. Eymard; T. Stojkovic; C. Wallgren-Pettersson; P. Carlier; D. Gomez Andres; B. Dore; E. Malfatti; N. Romero; R. Carlier

G.P.269 **CAP-disease not-related to *ACTA1, TPM2 or TPM3* genes**
M. Garibaldi; E. Malfatti; G. Brochier; J.M. Cuisset; C.A. Maurage; N. Monnier; B. Eymard; J. Laporte; M. Fardeau; N.B. Romero

G.P.270 **A large genomic rearrangement affecting TPM3 causing severe nemaline myopathy**
V.L. Lehtokari; K. Kiiski; K. Pelin; C. Wallgren-Pettersson

G.P.271 **Mutation update and genotype-phenotype correlations of novel and previously described mutations in TPM2 and TPM3 causing congenital myopathies**
M. Marttila; V.L. Lehtokari; S.B. Marston; T.A. Nyman; C. Barnerias; A.H. Beggs; E. Bertini; Ö. Ceyhan-Birsoy; P. Cintas; M. Gerard; B. Gilbert-Dussardier; J.S. Hogue; C. Longman; B. Eymard; M. Frydman; P.B. Kang; L. Klinge; H. Kolski; H. Lochmüller; L. Magy; V. Manel; M. Mayer; K.N. North; S. Peudenier-Robert; H. Pihko; F.J. Probst; R. Reisin; W. Stewart; A.L. Taratuto; M. de Visser; E. Wilichowski; J. Winer; K. Nowak; N.G. Laing; T.L. Winder; N. Monnier; N.F. Clarke; K. Pelin; M. Grönholm; C. Wallgren-Pettersson

G.P.272 **Novel deletions in TPM3 define a hypercontractile phenotype with marked congenital muscle stiffness: Expanding the spectrum of TPM3 related disease**
S. Donkervoort; M. Neu; J. Kirschner; M.L. Yang; S.B. Marston; M.A. Gibbons; Y. Hu; J.M. de Winter; C.A.C. Ottenheijm; A. Rutkowski; M. Krüger; E. McNamara; R. Ong; K. Nowak; N.F. Clarke; C.G. Bönnemann

G.P.273 **Nemaline myopathy 8 and KLHL40 in diseased and normal skeletal muscle**
G. Ravenscroft; E.J. Todd; K.S. Yau; C.A. Sewry; C.A. McLean; M.M. Ryan; R.J. Allcock; N.G. Laing

G.P.274 **Cold shock domain protein A - a novel nemaline myopathy-causing gene?**
J. Laitila; V.L. Lehtokari; K. Kiiski; C. Wallgren-Pettersson; K. Pelin

LGMD2; (G.P.275–289)

Facilitators: Isabelle Richard and Ieke Ginjaar

G.P.275 **Expanding the clinical and genetic spectrum of FKRP-related myopathies in Argentinean pediatric patients**
F. Lubieniecki; S. Monges; J. Mozzoni; A. Moresco; V. Aguerre; M.F. de Castro; N. Pozzo; E. Foncuberta; C. Bouchet Seraphin; N.B. Romero; A.L. Taratuto

G.P.276 **Slowly progressive motor and respiratory dysfunction resulting from FKRP mutations: A natural history study**
C.D. Crockett; C.M. Stephan; S.R.H. Mockler; K.M. Laubscher; B.M. Zimmerman; K.D. Mathews

G.P.277 **LGMD2I: Is there a relationship between clinical phenotype, morphological alterations and level of alpha-dystroglycan glycosylation in patients with the same FKRP genotype?**
S. Lindal; E. Stensland; M. Rasmussen; C. Jonsrud; V. Brox; A. Maisoon; Ø. Nilssen

G.P.278 **Facial dysmorphism in FKRP limb-girdle muscular dystrophy: About two cases**
A. Magot; S. Mercier; C. Bouchet Seraphin; J.M. Mussini; Y. Peron

G.P.279 **Efficient AAV-mediated transfer of FKRP in a new mouse model of Limb Girdle Muscular Dystrophy 2I**
E. Gicquel; I. Richard

G.P.280 **Preserved expression of truncated telethonin in a patient with LGMD2G**
V. Straub; N.P. Davies; R. Barresi; C. Morris; C. Pickthall; K. Bushby

G.P.281 **Detection of homozygous and compound heterozygous deletions in TRIM32 in LGMD patients analyzed by a combined strategy of CGH-array and Massive Parallel Sequencing**
J. Nectoux; R. de Cid; S. Baulande; F. Leturcq; J.A. Urtizberea; I. Penisson-Besnier; A. Nadaj Pakleza; C. Roudaut; A. Criqui; L. Orhant; D. Peyroulan; R. Ben Yaou; I. Nelson; M.C. Arné-Bes; P. Nitschke; M. Claustres; G. Bonne; N. Lévy; J. Chelly; I. Richard; M. Cossée

G.P.282 **A novel mutation in DNAJB6 causes a more severe phenotype and greater loss of anti-aggregation function**
P.H. Jonson; J. Palmio; J. Sarparanta; H. Luque; B. Udd

G.P.283 **A novel mutation in DNAJB6 gene causes a very severe early-onset LGMD1D disease**
J.M.M. Palmio; A. Evilä; P.H. Jonson; M. Auranen; S. Kiuru-Enari; H. Pihko; P. Hackman; B. Udd

G.P.284 **Dysferlinopathy caused by protein misfolding: The novel murine animal model Dysf-MMex38**
L. Heidt; M. Bader; S. Spuler; V. Schoewel

G.P.285 **Membrane and phospholipid binding properties of dysferlin**
J. Hofhuis; S. Thoms; L. Klinge

G.P.286 **Validation of *in silico* variation effect prediction tools in missense mutations of dysferlinopathy**
J.S. Park; S.J. Hwang; D.S. Kim; J.H. Shin

G.P.287 **Genetic and epigenetic determinants of low dysferlin expression in monocytes**
E. Gallardo; A. Ankala; Y. Nuñez-Alvarez; M. Hedge; J. Diaz-Manera; N. De Luna; A. Pastoret; M. Suelves; I. Illa

G.P.288 **Dysferlinopathy in Egypt: Clinical, Pathological and Genetic characteristics**
N.A. Fahmy; A. Abd Elhady; A. Abd El-Naser; S. Ashour; A. Etribi; I. Nonaka; N. Minami; N. Suzuki; T. Takahashi; M. Aoki

G.P.289 **Novel ANO5 KO Mouse Provides New Insight into LGMD2L Pathogenesis**
D. Griffin; E. Pozsgai; R. Johnson; W. Grose; K. Heller; J.R. Mendell; Z. Sahenk; L.R. Rodino-Klapac

Outcome measures 2; (T.P.35–48)

Facilitators: Yoram Nevo and Fiona Norwood

T.P.35 **Developing the SMA REACH UK database: A combined effort to improve standards of care and translational research in Spinal Muscular Atrophy**
D. Ramsey; M. Scoto; A. Mayhew; M. Main; I. Wilson; E. Mazzone; J. Montes; K. Bushby; R. Finkel; E. Mercuri; F. Muntoni

T.P.36 **Hammersmith Motor Function Scale and Upper Limb Module: 12 month correlation**
E. Mazzone; S. Sivo; M. Pane; C. Palermo; A. D'Amico; L. Fanelli; R. De Sanctis; E. Bertini; E. Mercuri

T.P.37 **Old measures and new analysis in non ambulant SMA patients**
E.S. Mazzone; A. Mayhew; M. Main; J. Montes; D. Ramsey; M. Scoto; M. Pane; K. Bushby; R. Finkel; F. Muntoni; E. Mercuri

T.P.38 **Initial validity and test-retest reliability of ACTIVE-seated (Ability Captured Through Interactive Video Evaluation-seated) as an upper extremity outcome in Duchenne muscular dystrophy**
L.P. Lowes; L.N. Alfano; K.M. Berry; H. Yin; I. Dvorchik; K.M. Flanigan; J.R. Mendell

T.P.39 **Psychometric properties of ACTIVLIM in 2-year cross sectional records of the Belgian Neuromuscular Disease Registry**
C.S. Batcho; P.Y.K. Van den Bergh; P. Van Damme; A. Roy; M. Penta; *. BNMDR Scientific Committee

T.P.40 **Clinical rating scale for head control: A pilot study for reliability and validity of the Turkish version**
O. Yilmaz; I. Alemdaroglu; S. Subasi; E. Necati; A.A. Karaduman; H. Topaloglu

T.P.41 **Upper limb strength and function in patients with glycogen storage disease type 3**
V. Decostre; A. Nadaj-Pakleza; G. Ollivier; A. Canal; P. Laforêt; J.Y. Hogrel

T.P.42 **Comparison of three scales of motor function in two subtypes of patients with congenital muscular dystrophy**
K.M. Meilleur; M.S. Jain; L.S. Hynan; C.Y. Shieh; E. Kim; M. Waite; M. McGuire; C. Fiorini; A. Glanzman; M. Main; K. Rose; T. Duong; M.M. Linton; I.C. Arveson; C. Nichols; K. Yang; M. Smith; S. Donkervoort; M.E. Leach; A.R. Foley; J. Collins; F. Muntoni; A. Rutkowski; C.G. Bönnemann

T.P.43 **Effects of proximal and distal muscle weakness on performance in neuromuscular diseases**
I. Alemdaroglu; P. Kaya; O. Yilmaz; A.A. Karaduman; H. Topaloglu

T.P.44 **Influence of a two-year steroid treatment on body composition as measured by Dual X-Ray Absorptiometry in boys with Duchenne Muscular Dystrophy**
C. Vuillerot; P. Braillon; S. Fontaine-Carbonnel; P. Rippert; E. Andre; J. Iwaz; I. Poirot; C. Berard

T.P.45 **A new index of muscle development and disease progression for the pediatric patients with Duchenne muscular dystrophy, using bioelectrical impedance analysis; An Observation Study**
T. Uchiyama; A. Hattori; T. Sato; T. Murakami; K. Ishigaki; T. Nakayama; S. Kuru

T.P.46 **A new index of muscle development and disease progression for the pediatric patients with Fukuyama congenital muscular dystrophy, using bioelectrical impedance analysis; an observation study**
T. Murakami; T. Uchiyama; T. Sato; K. Ishigaki; T. Nakayama; S. Kuru

T.P.47 **A new index of muscle development for healthy children using bioelectrical impedance analysis**
T. Uchiyama; T. Nakayama; A. Hattori; S. Kuru

T.P.48 **Acoustic radiation force impulse imaging for the longitudinal assessment of muscle tissue stiffness in collagen 6 myopathy and LAMA2 related muscular dystrophy**
J. Dastgir; C. Vuillerot; D. Nguyen; K. Yang; S. Auh; S. Donkervoort; K. Meilleur; M. Leach; M. Jain; A. Rutkowski; C. Bönnemann

Neuropathies; (G.P.290–301)

Facilitators: Kristl Claeys and Hannes Vogel

G.P.290 **Prefrontal involvement related to cognitive impairment in progressive muscular atrophy**
J. Raaphorst; M.J. van Tol; P.F.C. de Groot; E. Altena; Y.D. van der Werf; C.B. Majoie; A.J. van der Kooi; L.H. van den Berg; B.A. Schmand; M. de Visser; D.J. Veltman

G.P.291 **Peripheral nerve and spinal root ultrasonography in amyotrophic lateral sclerosis**
C. Watanabe; T. Makino; R. Kumano; M. Higaki; H. Toji

G.P.292 **Nerve ultrasonography and neurophysiological study in amyotrophic lateral sclerosis to assess distal motor dysfunction of median nerve**
R. Kumano; T. Makino; M. Higaki; H. Toji; C. Watanabe

G.P.293 **Peripheral nerve conduction abnormalities in amyotrophic lateral sclerosis**
T. Makino; C. Watanabe; R. Kumano; M. Higaki; H. Toji

G.P.294 **Charcot-Marie-Tooth disease type 1A presenting as muscle hypertrophy and muscle cramps**
E. Brusse; J.C. Perumpillichira

G.P.295 **Relationship between ankle instability and disability in children with Charcot-Marie-Tooth disease**
A.D. Sman; M. Mandarakas; M. Menezes; J. Burns

G.P.296 **Severe, early onset Charcot-Marie-Tooth disease with rare presentations**
S.W. Yum; L. Medne; T. Estilow; A. Glanzman; D. DiVito; C.G. Bönnemann

G.P.297 **Supramaximally stimulated complex A-waves are helpful to distinguish lower motor neurone disease from demyelinating neuropathies**
G. Remiche; N. Mavroudakis

G.P.298 **A case of bilateral neuralgic amyotrophy combined with phrenic nerve palsy**
J.M. Park; J.S. Jeon; J.S. Park

G.P.299 **Novel use of eculizumab in a patient with Guillain-Barré syndrome**
D. Ram; A. Sutherland; S. Hughes; G. Vassallo

G.P.300 **Pandysautonomia: A rare variant of Guillain-Barre syndrome**
M. Güngör; P. Esmeray; İ. Öncel; H. Topaloglu

G.P.301 **Intraneural Perineurioma: A Rare Cause of Monomelic Weakness**
D. Ram; C. Duff; G. McCullagh

Psychosocial approaches; (G.P.302–310)

Facilitators: Ulrike Schara and Jes Rahbek

G.P.302 **Perceived barriers and facilitators of participation in clinical trials for Duchenne and Becker muscular dystrophy**
H.L. Peay; H. Scharff; B.B. Biesecker; B.S. Wilfond; J. Johnson; D.M. Escolar; J. Bowie; K. Nagaraju; J. Piacentino; A. Tibben

G.P.303 **Influences on parental decision making about participation in Duchenne and Becker muscular dystrophy clinical trials**
H.L. Peay; H. Scharff; B.B. Biesecker; B.S. Wilfond; J. Johnson; D.M. Escolar; K. Nagaraju; J. Piacentino; J. Bowie; A. Tibben

G.P.304 **Living conditions and quality of life in adults with Duchenne muscular dystrophy – A Danish survey**
A. Madsen; J. Rahbek; B. Werge; J. Marquardt; O. Gredal; B.F. Steffensen

G.P.305 **Pain in adolescents with spinal muscular atrophy and Duchenne and Becker muscular dystrophy**
C. Lager; A.K. Kroksmark

G.P.306 **Pediatric Neuromuscular Diseases Patient Profiles from Family Perspective in Turkey**
O. Yilmaz; A.A. Karaduman; S. Subasi; S. Serel; H. Topaloglu

G.P.307 **Skype and psychoanalysis: An approach to circumvent patients locomotion difficulties**
J. Forbes; L. Lise; T. Genesini; C. Riolfi; E. Macedo; G. Achôa; D. Rüdiger; L. Fachinetto; M. Neves; A. Mouzat; R.C.M. Pavanello; M. Zatz

G.P.308 **Perception of efficacy in adult patients affected by Spinal Muscular Atrophy (SMA) treated with salbutamol**
M.B. Pasanisi; A.M. Giovannetti; C. Bussolino; A. Campanella; M. Leonardi; L. Morandi

G.P.309 **Exploring the motivations for clinic attendance in Myotonic dystrophy (DM1) and Huntington's disease (HD)**
K.A. LaDonna; S.L. Ray; S.L. Venance

G.P.310 **The cognitive profile of ALS: Update of a meta-analysis**
E. Beeldman; J. Raaphorst; R.J. De Haan; M. De Visser; B.A. Schmand

CMD Alpha-dystroglycan; (G.P.311–321)

Facilitators: Tatsushi Toda and Ikuya Nonaka

G.P.311 **A new homozygous ISPD mutation is associated with either early limb-girdle or congenital muscular dystrophy within the same family depending on different levels of alpha-dystroglycan glycosylation**
G. Baranello; L. Morandi; S. Sansanelli; P. Savadori; S. Saredi; C. Pantaleoni; P. Balestri; A. Malandrini; M.T. Arnoldi; L. Chiapparini; M. Mora

G.P.312 **Intrafamilial variability in *GMPPB* associated alpha-dystroglycanopathy and broadening of the clinical phenotype**
D.X. Bharucha-Goebel; E. Neil; S. Donkervoort; S. Moore; T. Winder; J. Dastgir; S. Iannaccone; C.G. Bönnemann

G.P.313 **Intrafamilial heterogeneity in an alpha-dystroglycanopathy due to GDP-Mannose Pyrophosphorylase B (GMPPB) mutations**
M. Bertoli; T. Evangelista; A. Sarkozy; A. Schaefer; P. Goldsmith; R. Barresi; V. Straub; F. Muntoni; K. Bushby; H. Lochmuller

G.P.314 **Alpha-dystroglycanopathy: Two new patients with GMPPB mutations and a mild limb-girdle phenotype**
M. Sframeli; A. Sarkozy; C. Longman; L. Feng; S. Robb; A.Y. Manzur; R. Mein; M. Yau; R. Barresi; R. Phadke; C. Sewry; F. Muntoni

G.P.315 **Cases of normal to mildly elevated creatine kinase in muscle-eye-brain disease patients and delay in diagnosis**
S. Joseph; C. Longman; F. Muntoni; I. Horrocks

G.P.316 **Effectiveness of levetiracetam for seizures in patients with Fukuyama congenital muscular dystrophy**
T. Sato; K. Ishigaki; M. Shichiji; T. Saito; T. Murakami; K. Saito; M. Osawa; S. Nagata

G.P.317 **Renal dysfunction in patients with Fukuyama congenital muscular dystrophy**
K. Ishigaki; T. Murakami; T. Sato; K. Ishiguro; M. Funatsuka; I. Kato; K. Saito; M. Osawa; S. Nagata

G.P.318 **Natural history of motor function in patients with Fukuyama congenital muscular dystrophy**
K. Ishiguro; K. Ishigaki; T. Sato; T. Murakami; K. Saito; M. Osawa; S. Nagata

G.P.319 **Central nervous system involvement in the animal model of Congenital Muscular Dystrophy 1D (Large)**
C.M. Comim; B.P. Mendonça; D. Dominguini; M. Vainzof; E.L. Streck; F. Dal-Pizzol; J. Quevedo

G.P.320 **Phenotypic heterogeneity of the c.919T > A FKRP gene mutation in humans and a mouse model**
E. Rivas; J. Vissing; M. Scoto; M. Fernandez-Fuente; T. Voit; F. Muntoni; S. Brown

G.P.321 **A new monoclonal antibody against human alpha-dystroglycan has potential diagnostic applications**
E.L. Humphrey; E. Lacey; L.T. Le; F. Sciandra; C. Morris; J.E. Hewitt; I. Holt; A. Brancaccio; R. Barresi; C.A. Sewry; S.C. Brown; G.E. Morris

18:00–20:00 **Genzyme Industry Symposium** – Langenbeck-Virchow-Haus (Foyer & Lecture Hall)

Friday 10 October 2014

09:00–10:00 **Advances in Therapy of Neuromuscular Disorders;** Invited lectures (T.I.1–2) – Langenbeck-Virchow-Haus
Chairpersons: Ronald D Cohn and Ichizo Nishino

T.I.1 **The challenge of making therapies happen for neuromuscular diseases**
T. Voit

T.I.2 **Therapeutic efforts in GNE myopathy**
Z. Argov

10:00–10:30 **Morning Tea and Coffee** – Langenbeck-Virchow-Haus (Foyer)

10:30–12:30 **Treatment approaches for neurogenic disorders;** Invited lecture (G.O.15–22) – Langenbeck-Virchow-Haus
Chairpersons: Anders Oldfors and Douglas Biggar

G.O.15 **Effect of early and late restoration of SMN in SMA animal models and identification of genes downstream of SMN**
A. Burghes; D. Arnold; S. Duque; V. McGovern; C. Ruhno; C. Iyer; K. Meyer; B. Kaspar

G.O.16 **Reliability and validity of the ACTIVE-mini (Ability Captured Through Interactive Video Evaluation-mini) to quantify infant movement**
L.P. Lowes; L.N. Alfano; K.M. Berry; S. Wolock; R.W. Rumpf; W. Ray; A. Meyer; J. Jackson; A. Firmalan; K.M. Flanigan; B. Kaspar; J.R. Mendell

G.O.17 **Results of a Phase 2 Open-Label Study of ISIS-SMNRx in Patients with Infantile (Type 1) Spinal Muscular Atrophy**
R.S. Finkel; J. Day; C. Chiriboga; J. Vasjar; D. Cook; K. Watson; S. Paulose; L. McMillian; R. Cruz; J. Montes; D.C. De Vivo; M. Yamashita; D. McGuire; K. Alexander; D. Norris; C.F. Bennett; K. Bishop

G.O.18 **Results of a Phase 2 Study of ISIS-SMNRx in Children with Spinal Muscular Atrophy**
B. Darras; C. Chiriboga; K. Swoboda; S. Iannaccone; J. Montes; D. Castro; N. Holuba; N. Raush; N. Visyak; S. Dunaway; D. Trussell; A. Pasternak; L. Neilson; D. De Vivo; D. McGuire; D. Norris; K. Alexander; C. F. Bennett; K. Bishop

G.O.19 **Results of a Phase II Study to Assess Safety and Efficacy of Olesoxime (TRO19622) in 3-25 Years Old Spinal Muscular Atrophy Patients**
E. Dessaud; C. André; B. Scherrer; P. Berna; R. Pruss; V. Cuvier; W. Hauke; C. Bruno; B. Chabrol; G. Comi; J.M. Cuisset; N. Deconinck; N. Goemans; B. Estournet; S. Fontaine-Carbonel; K. Gorni; J. Kirschner; A. Lusakowska; H. Lochmuller; M. Mayer; E. Mercuri; W. Müller-Felber; F. Muntoni; F. Rivier; H. Roper; U. Schara; L. Van den Berg; G. Vita; M. Walter; E. Bertini

G.O.20 **AAV-mediated overexpression of Ubiquilin2 mimics ALS and ALS with dementia in naive mice**
M.G. Biferi; C. Bos; Y. Tanguy; S. Alonso-Martin; B. Artegiani; M. Roda; M. Cohen-Tannoudji; A. Ferry; S. Camerini; M. Crescenzi; F. Calegari; F. Relaix; M. Barkats

G.O.21 **Relapsing immune mediated polyneuropathy, strokes and chronic haemolysis due to inherited CD59 deficiency**
B. Sayınbatur; J. Maluenda; C. Temuçin; B. Tavil; M. Çetin; K. Karlı-Oguz; I. Gut; G. Haliloglu; J. Melki; H. Topaloglu

G.O.22 **Reducing dynamin 2 rescues centronuclear myopathy**
B.S. Cowling; T. Chevremont; I. Prokic; H. Tasfaout; C. Kretz; A. Ferry; C. Coirault; V. Laugel; N.B. Romero; J. Laporte

12:30–14:00 **Lunch packages** – Langenbeck-Virchow-Haus (Foyer)

14:00 **Free Afternoon (Group boat trip)**

19:30–24:00 **Congress Dinner –** Ludwig Loewe Höfe, Loewe Saal

Saturday 11 October 2014

09:00–10:30 **Treatment approaches in the clinic;** Oral Presentations (G.O.23–28) – Langenbeck-Virchow-Haus
Chairpersons: Norma Beatriz Romero and Eugenio Mercuri

G.O.23 **Drisapersen: An overview of the clinical programme to date in Duchenne Muscular Dystrophy (DMD)**
C. McDonald; E. Mercuri; N. Goemans; T. Voit; R. Wilson; C. Wardell; G. Campion

G.O.24 **Eteplirsen in Duchenne Muscular Dystrophy (DMD): 3 year update on Six-Minute Walk Test (6MWT) and Safety**
J.R. Mendell; L. Rodino-Klapac; Z. Sahenk; K. Roush; L. Bird; L.P. Lowes; L. Alfano; A.M. Gomez; S. Lewis; V. Malik; K. Shontz; K.M. Flanigan; C. Shilling; P. Sazani; J. Saoud; P. Duda; E. Kaye

G.O.25 **Follistatin Gene Therapy for Becker Muscular Dystrophy**
J.R. Mendell; Z. Sahenk; L.R. Rodino-Klapac; K.R. Clark; K.R. Lewis; K. Shontz; S. Al-Zaidy; L. Alfano; L.P. Lowes; K. Berry; V. Malik; C.J. Shilling; X.Q. Rosales; C.M. Walker; K.M. Flanigan; M. Hogan; B.K. Kaspar

G.O.26 **Human α7 integrin gene (ITGA7) delivered by adeno-associated virus reverses the phenotype of the double knock out (DKO) mouse devoid of dystrophin and utrophin**
K.N. Heller; C.L. Montgomery; K.M. Shontz; K.R. Clark; J.R. Mendell; L.R. Rodino-Klapac

G.O.27 **Randomized controlled phase 2 study of GNE myopathy subjects treated with placebo, 6g or 3g doses of extended-release sialic acid (SA-ER) and an extension study at a higher 12 g dose of combination extended and immediate release sialic acid (SA)**
H. Lau; Z. Argov; Y. Caraco; A. Pestronk; P. Shieh; A.M. Skrinar; J. Mayhew; J. Martinisi; E. Kakkis

G.O.28 **Sporadic late onset nemaline myopathy with MGUS: Long term follow-up after melphalan and autologous stem cell transplantation**
N.C. Voermans; O. Benveniste; M. Minnema; H. Lokhorst; M. Lammens; W. Meersseman; M. Delforge; T. Kuntzer; J. Novy; T. Pabst; F. Bouhour; N. Romero; V. Leblond; P. Van den Bergh;

10:30–11:00 **Morning Tea and Coffee** – Langenbeck-Virchow-Haus (Foyer)

11:00–12:30 **Poster Highlights** – Langenbeck-Virchow-Haus

Chairpersons: Beril Talim and Vincenzo Nigro

12:30–13:30 **WMS General Assembly** – Langenbeck-Virchow-Haus (Lecture Hall)

13:30–14:30 **Lunch** – Langenbeck-Virchow-Haus (Foyer)

14:30–16:00 **Late Breaking Session** – Langenbeck-Virchow-Haus

Chairpersons: Nigel Laing and Gisèle Bonne

16:00 **Prize giving and Welcome to the 20th WMS Congress** – Langenbeck-Virchow-Haus
Handover of the WMS flag and closing of the Congress