20th WMS Congress – 2015 – Program

Wednesday 30 September 2015

12:30–16:00		**WMS Executive Board Meeting** – Thistle Hotel (Keats room in executive suite)
14:00–18:00		**Registration** – Brighton Dome foyer
		Setting up of posters – Brighton Dome foyer
18:00–18:45		**Opening ceremony** – Concert Hall Dolphins and cows, Neal Maskrey, National Institute for Health and Care Excellence (NICE)
18:45–21:00		**Welcome reception** – Brighton Museum – Corn Exchange, café and mezzanine

Thursday 1 October 2015

07:45		**Conference desk opens** – Brighton Dome foyer
08:15–08:40		**20 years of WMS – Victor Dubowitz** – Concert Hall
08:40–09:00		**Congress opening – Message from the President Victor Dubowitz** – Concert Hall
09:00–10:30		**Muscle metabolism in health and disease;** Invited lectures (M.I.1–3) *Chairpersons: Victor Dubowitz and Francesco Muntoni*
	M.I.1	**Exercise treatment in muscle disorders** J. Vissing
	M.I.2	**Exercise and mitochondrial function in health and disease** R. Haller
	M.I.3	**Genetic modifiers of muscle: Studies of college students and Duchenne muscular dystrophy** E. Hoffman; P. Thompson; C. McDonald; H. Gordish-Dressman; L. Bello; CINRG Investigators; FAMuSS Investigators; E. Pegoraro
10:30–11:00		**Morning refreshments, exhibition and posters** – Corn Exchange, café and mezzanine
11:00–11:30		**Muscle metabolism in health and disease;** Invited lectures (M.I.4) – Concert Hall *Chairpersons: Ros Quinlivan and Vincenzo Nigro*
	M.I.4	**Regulation of skeletal muscle metabolic and angiogenic properties by nuclear hormone receptors: implications for skeletal muscle regeneration** P. Sfyri; V. Narkar; A. Matsakas
11:30–13:00		**Neuromuscular diseases;** Oral presentations (G.O.1–6) – Concert Hall *Chairpersons: Ros Quinlivan and Vincenzo Nigro*
	G.O.1	**Muscle disorders of glycogen metabolism associated with *GYG1* and *RBCK1* deficiency** A. Oldfors
	G.O.2	**A gene for speed: the influence of ACTN3 on muscle performance in health and disease** P. Houweling; J. Seto; F. Garton; K. Quinlan; S. Head; K. North

G.O.3 **Dysregulated mitophagy and mitochondrial transport in sensori-motor neuropathy due to "Dominant Optic Atrophy" plus with OPA1 (Optic Atrophy 1) mutations**
C. Liao; A. Diot; N. Ashley; K. Morten; C. Fratter; I. Moroni; S. Bianchi; C. Lamperti; E. Dombi; S. Downes; K. Sitarz; P. Yu-Wai-Man; A. Simon; M. Reilly; T. Enver; F. Iborra; M. Votruba; H. Mortiboys; M. Zeviani; J. Poulton

G.O.4 **Autosomal recessive mutations of GPR126 are responsible for severe arthrogryposis multiplex congenita**
G. Ravenscroft; F. Nolent; S. Rajagopalan; A. Meireles; K. Paavola; D. Gaillard; E. Alanio; M. Buckland; S. Arbuckle; M. Krivanek; J. Maluenda; S. Pannell; R. Gooding; R. Ong; R. Allcock; F. Kok; W. Talbot; J. Melki; N. Laing

G.O.5 **A premature stop codon in MYO18B is associated with severe nemaline myopathy with cardiomyopathy**
E. Malfatti; J. Böhm; E. Lacène; N. Romero; J. Laporte

G.O.6 **LRP4 myasthenia. Investigation of a second kinship reveals impaired development and maintenance of the neuromuscular junction**
D. Selcen; B. Ohkawara; X. Shen; K. McEvoy; K. Ohno; A. Engel

13:00–14:30 **Lunch, exhibition and posters** – Corn Exchange, café and mezzanine

14:30–16:00 **Guided poster discussion session 1: parallel sessions (G.P.1–111)** – Corn Exchange

Pompe Disease (G.P.1–15)
Facilitators: Anders Oldfors and Pascal Laforêt

G.P.1 **Cytokine and chemokine profiling shows rapid activation of the immune system following enzyme replacement therapy in Pompe disease**
E. Masat; P. Laforêt; D. Amelin; P. Veron; B. Perniconi; N. Taouagh; O. Benveniste; F. Mingozzi

G.P.2 **Vaginal birth in a patient with Pompe disease**
B. Perniconi; D. Vauthier-Brouzes; C. Morelot-Panzini; M. Dommergues; J. Nizard; N. Taouagh; L. Servais; P. Laforêt

G.P.3 **The importance of screening for late onset Pompe disease in neuromuscular disorders**
M. Matsui; T. Saitou; T. Matsumura; K. Toyooka; K. Inoue; H. Fujimura; S. Sakoda

G.P.4 **Physiotherapy in late-onset Pompe disease: a 3 year review of exercise and muscle strength**
S. Dando

G.P.5 **Blood film examination for vacuolated and PAS-positive lymphocytes as diagnostic screening test for patients with late onset Pompe disease (LOPD)**
A. Toscano; D. Parisi; F. Montagnese; M. Marino; A. Migliorato; A. Ciranni; C. Rodolico; O. Musumeci

G.P.6 **Whole body muscle MRI correlates with muscle function in patients with adult onset Pompe disease**
J. Díaz-Manera; S. Segovia; J. Llauger; I. Belmonte; G. Suárez-Cuartin; M. Mayos; A. Alejaldre; S. Figueroa-Bonaparte; I. Pedrosa; L. Querol; R. Rojas-García; E. Gallardo; I. Illa; Spanish Pompe Study Group

G.P.7 **PGM1 deficiency – A heterogeneous myopathy with opportunities for treatment**
N. Voermans; N. Preisler; K. Madsen; M. Janssen; B. Kusters; D. Maas; J. Groothuis; J. Vissing; B. Engelen; D. Lefeber

G.P.8 **An international, phase 3, switchover study of reveglucosidase alfa (BMN 701) in subjects with late-onset Pompe disease**
B. Schoser; B. Byrne; F. Eyskens; T. Hiwot; D. Hughes; J. Kissel; E. Mengel; T. Mozaffar; A. Pestronk; M. Roberts; K. Sivakumar; J. Statland; P. Young; C. Heusner; W. Dummer

G.P.9 **The impact of enzyme replacement therapy on the progression of the Pompe disease**
M. De Antonio; D. Hamroun; B. Perniconi; N. Taouagh; E. Salort-Campana; S. Sacconi; F. Zagnoli; P. Laforêt and French Pompe Study Group

G.P.10 **A higher dose of alglucosidase alfa in classic infantile Pompe disease positively affects ventilator-free survival and motor outcome: an open-label single-center study**
C. van Gelder; E. Poelman; I. Plug; M. Hoogeveen-Westerveld; E. Kuperus; N. van der Beek; A. Reuser; A. van der Ploeg

G.P.11 **Characteristic skeletal muscle imaging patterns in Japanese patients with Pompe disease**
K. Ishigaki; H. Kobayashi; H. Sugie; T. Fukuda; T. Murakami; T. Sato; K. Ishiguro; M. Shichiji; S. Nagata; M. Osawa; Y. Eto; I. Nonaka

G.P.12 **Childhood Pompe disease: clinical spectrum and genotype in 31 children**
F. van Spronsen; J. Jaeken; M. Rubio-Gozalbo; J. van der Meijden; M. Willemsen; M. Baethmann; R. Lachmann; C. van Capelle; E. Mengel; T. Voit; H. Michelakakis; J. van den Hout; A. Reuser; M. Kroos; A. van der Ploeg

G.P.13 **Intracranial arterial abnormalities in patients with late onset Pompe disease**
A. Toscano; F. Montagnese; F. Granata; C. Rodolico; S. Mondello; M. Cucinotta; A. Ciranni; M. Longo; O. Musumeci

G.P.14 **Lung function tests (MIP, MEP, FVC, VC) predict ventilation and wheelchair use in late-onset Pompe disease**
M. Roberts; T. Mozaffar; P. Young; E. Johnson; A. Quartel; K. Berger

G.P.15 **Identification and characterization of aberrant splicing in Pompe disease using a genetic approach**
W. Pijnappel; A. Bergsma; A. van der Ploeg; M. Hoogeveen-Westerveld; D. Halley; M. Kroos

SMA (G.P.16–33)
Facilitators: Richard Finkel and Susan Iannaccone

G.P.16 **Proactive, reactive and palliative approaches to care for pediatric patients with SMA1: a retrospective analysis**
K. Shellenbarger; B. Wong; I. Rybalsky; H. Sawnani; R. Amin; B. Godshall; P. Sturm; T. Inge; N. Weidner

G.P.17 **The experiences of individuals and families affected by spinal muscular atrophy (SMA) and their views on living with SMA**
Y. Qian; S. McGraw; J. Henne; J. Jarecki; K. Hobby; W. Yeh

G.P.18 **Parents' experiences of having a child with spinal muscular atrophy type 1: informing clinical practice**
R. Forbes; B. McClaren; M. Sahhar; M. Ryan; E. Higgs

G.P.19 **Perspective from spinal muscular atrophy families: care of a child with tracheostomy and home mechanical ventilatory support**
M. Tanyildiz; H. Topaloğlu; I. Oncel; B. Bayrakci; G. Haliloglu

G.P.20 **Spontaneous breathing pattern in children with spinal muscular atrophy: correlation with motor function assessment**
G. Baranello; M. Arnoldi; C. Bussolino; C. Mastella; A. Aliverti; A. Lo Mauro

G.P.21 **Response of biomarkers to treatment in mouse models of spinal muscular atrophy and response of SMA mice to later treatment**
A. Burghes; W. Arnold; V. McGovern; S. Duque; J. Li; C. Iyer; K. Corlett; K. von Herrmann; P. Zaworski; K. Chen; S. Kolb; S. Paushkin; D. Kobayashi; S. Rutkove

G.P.22 **Generation and rescue of intermediate SMA mice by low-dose morpholino antisense oligomer**
H. Zhou; J. Meng; E. Marrosu; N. Janghra; J. Morgan; F. Muntoni

G.P.23 **Microvascular abnormality in spinal muscular atrophy and its response to antisense oligonucleotide therapy**
H. Zhou; H. Ying; M. Scoto; P. Brogan; S. Parson; F. Muntoni

G.P.24 **A novel evaluation method of survival motor neuron protein as a biomarker of spinal muscular atrophy**
M. Arakawa; R. Arakawa; R. Aoki; A. Nomoto; K. Saito; M. Shibasaki

G.P.25 **Dysregulation of microRNAs in spinal muscular atrophy and the response to antisense oligonucleotide therapy**
H. Zhou; F. Catapano; I. Zaharieva; M. Scoto; J. Morgan; F. Muntoni

G.P.26 **Repetitive nerve stimulation in spinal muscular atrophy**
C. Preterre; A. Magot; F. Geneau Bodis; S. Mercier; G. Fayet; L. Servais; Y. Péréon

G.P.27 **Type-specific selectivity pattern of skeletal muscle images in spinal muscular atrophy**
M. Inoue; A. Ishiyama; H. Komaki; E. Takeshita; Y. Shimizu-Motohashi; T. Saito; E. Nakagawa; K. Sugai; N. Minami; Y. Goto; M. Sasaki

G.P.28 **Revised Hammersmith scale for spinal muscular atrophy: Inter & intra-rater reliability and content validity from a patient perspective**
D. Ramsey; M. Scoto; A. Mayhew; G. Ramdharry; A. Wallace; F. Muntoni

G.P.29 **Revised Hammersmith scale for spinal muscular atrophy**
D. Ramsey; M. Scoto; A. Mayhew; M. Main; I. Wilson; E. Mazzone; J. Montes; S. Dunaway; A. Pasternak; R. Salazar; K. Bushby; R. Finkel; E. Mercuri; F. Muntoni

G.P.30 **A European prospective study of the natural history of patients with type 2 and 3 spinal muscular atrophy**
M. Annoussamy; A. Chabanon; A. Phelep; J. Hogrel; P. Carlier; E. Fournier; R. Hermosilla; G. Ramey; C. Czech; L. Lowes; Y. Péréon; C. Cances; J. Cuisset; V. Laugel; C. Vuillerot; A. Daron; N. Goemans; U. Schara; T. Voit; L. Servais

G.P.31 **Low ovarian reserve in girls with autosomal-recessive proximal spinal muscular atrophies type I–III**
H. Koelbel; B. Hauffa; A. Boukidis; S. Lutz; A. Della-Marina; U. Schara

G.P.32 **Visual-cognitive and motor skills in children with type I spinal muscle atrophy**
M. Voos; G. Polido; A. Barbosa; F. Favero; F. Caromano

G.P.33 **Clinical and pathological features in SMA patients confirmed by genetic analysis**
J. Lee; S. Kim; Y. Choi

DMD – gene and imaging (G.P.34–53)
Facilitators: Haluk Topaloğlu and Valeria Ricotti

G.P.34 **Audit of DEXA bone density scans in boys with Duchenne muscular dystrophy who have sustained fractures**
T. Willis; N. Emery; R. Kulshrestha

G.P.35 **The evaluation of bone age in patients with Duchenne muscular dystrophy on long-term glucocorticoid treatment**
E. Annexstad; J. Westvik; A. Myhre; J. Bollerslev; I. Holm; M. Rasmussen

G.P.36 **Muscle ultrasound in Duchenne muscular dystrophy: Useful tool to monitor progression of early disease**
P. Karachunski; J. Dalton; J. Marsh

G.P.37 **MRI-derived apparent fat fractions in the thigh muscles of ambulant boys with Duchenne muscular dystrophy in a double blind clinical trial of drisapersen**
C. Bishop; R. Newbould; A. Quartel; A. Hall; G. Campion

G.P.38 **Longitudinal quantitative muscle magnetic resonance imaging (qMRI) in five boys with Duchenne muscular dystrophy (DMD), on and off treatment with drisapersen**
M. Hooijmans; B. Wokke; N. Goemans; G. Campion; J. Verschuuren; H. Kan; E. Niks

G.P.39 **Longitudinal quantitative muscle MRI in 5 Duchenne boys on and off drisapersen treatment**
M. Hooijmans; B. Wokke; N. Goemans; G. Campion; J. Verschuuren; H. Kan; E. Niks

G.P.40 **Magnetic resonance imaging of the arm and shoulder of boys with DMD**
R. Willcocks; S. Forbes; W. Triplett; D. Lott; C. Senesac; T. Nicholson; H. Arora; G. Walter; K. Vandenborne

G.P.41 **Long-term follow up of patients with dystrophin deficiency and isolated hyperCKemia**
P. Marti; N. Muelas; F. Mayordomo; I. Azorin; S. Blanch; C. Millan; T. Jaijo; J. Vilchez

G.P.42 **Idiopathic intracranial hypertension requiring a lumbar-peritoneal shunt in a child with Duchenne muscular dystrophy on daily steroids**
T. Willis; R. Kulshrestha; Z. Alhaswani

G.P.43 **Trends of steroid therapy for Duchenne muscular dystrophy in Japan**
F. Takeuchi; H. Komaki; H. Nakamura; N. Yonemoto; K. Kashiwabara; E. Kimura; S. Takeda

G.P.44 **Addressing discrepancies in care with the certified Duchenne care center program**
K. Kinnett; J. Mendel; J. Dowling; E. Wagner; L. Cripe; S. Apkon; R. Shell; R. Finkel; S. Nelson; D. Matthews; J. Porter; P. Furlong

G.P.45 **Cautionary tale in Duchenne muscular dystrophy – Opioids in neuromuscular disorders**
C. Marini Bettolo; M. Guglieri; H. van Ruiten; V. Straub; K. Bushby; H. Lochmüller

G.P.46 **A pilot study using solution-focused coaching for health promotion in children and young people with Duchenne muscular dystrophy**
A. McPherson; L. McAdam; H. Schwellnus; S. Keenan; E. Biddis; K. English; A. DeFinney

G.P.47 **Duchenne muscular dystrophy caused by a DMD nonsense mutation transmitted from a grandfather with somatic mosaicism**
E. Ulm; M. Walker; B. Wong

G.P.48 **Correlation of respiratory function parameters in 10–18 year old patients with Duchenne muscular dystrophy**
C. Rummey; T. Meier; M. Leinonen; C. McDonald; G. Buyse

G.P.49 **The use of a hand-held device (ASMA-1) for home-based monitoring of respiratory function changes in pediatric and adolescent patients with Duchenne muscular dystrophy**
T. Meier; C. Rummey; M. Leinonen; T. Voit; U. Schara; C. Straathof; M. D'Angelo; G. Bernert; J. Cuisset; R. Finkel; N. Goemans; C. McDonald; G. Buyse

G.P.50 **Transduce: Multidisciplinary out-patient transition in adolescents and young adults with Duchenne muscular dystrophy**
J. Groothuis; H. Hijdra; C. Erasmus; J. Raaphorst; I. de Groot

G.P.51 **Impact of three decades of improvement in standards of care for Duchenne muscular dystrophy**
C. Jimenez Moreno; M. Eagle; A. Mayhew; M. James; V. Straub; K. Bushby

G.P.52 **Improving the care of New Zealanders with Duchenne muscular dystrophy**
M. Rodrigues; R. Patel; R. Roxburgh

G.P.53 **Is functional dependence of Duchenne muscular dystrophy patients determinant of the quality of life and burden of their caregivers?**
M. Soares de Moura; H. Wutski; M. Voos; M. Resende; U. Reed; R. Hasue

Mitochondrial and lipid disorders (G.P.54–69)
Facilitators: Mike Hanna and Antonio Toscano

G.P.54 **Chemotherapy-induced mitochondrial respiratory dysfunction, oxidant production and death in healthy skeletal muscle C2C12 myoblast and myotube models**
B. Cheregi; C. Timpani; K. Nurgali; A. Hayes; E. Rybalka

G.P.55 **Acylcarnitine profiles in mitochondrial myopathies**
C. Vissing; M. Christensen; M. Dunø; F. Wibrand; J. Olesen; J. Vissing

G.P.56 **Reduced fatty acid oxidation capacity during exercise in patients with carnitine transporter deficiency**
K. Madsen; N. Preisler; J. Rasmussen; G. Petersen; J. Olesen; A. Lund; J. Vissing

G.P.57 **Exploration of muscle metabolism during exercise and muscle imaging in late-onset form of multiple Acyl-coA dehydrogenase deficiency (MADD)**
S. Souvannanorath; S. Hatem; M. Becquemin; N. Zraik; R. Bittar; O. Rigal; R. Carlier; P. Carlier; P. Laforêt

G.P.58 **Not motoneuron disease – A case of late-onset multiple acyl-CoA dehydrogenase deficiency**
C. Merkel; N. Boy; K. Kappes-Horn; J. Reimann

G.P.59 **Gene mutation screening using whole exome sequencing in lipid storage myopathy**
K. Takayama; S. Mitsuhashi; I. Nonaka; S. Noguchi; I. Nishino

G.P.60 **Mutations in iron-sulfur cluster assembly gene IBA57 cause progressive cavitating leukoencephalopathy**
A. Ishiyama; C. Sakai; Y. Endo; S. Mitsuhashi; S. Noguchi; Y. Matsushima; Y. Hayashi; H. Komaki; K. Sugai; M. Sasaki; I. Nonaka; Y. Goto; I. Nishino

G.P.61 **The value of the MOX accelerometer in measuring daily physical activity in children with mitochondrial disease**
S. Koene; I. Dirks; E. van Mierlo; A. Janssen; K. Meijer; H. Essers; J. Smeitink; A. Bergsma; I. de Groot

G.P.62 **MitoScale: A pre-diagnostic screen for the likelihood of mitochondrial disease in adult patients**
C. Liang; F. Edema-Hildebrand; C. Sue

G.P.63 **Musculoskeletal function in children with Barth syndrome**
C. Steward; K. Vijayakumar; S. Love; B. Toms; A. Majumdar; S. Badger; D. Goodman; K. Kurian; L. Markham; L. Jones

G.P.64 **Immunohistochemical detection of HIF1-alpha distribution in muscle of children with different neuromuscular disorders**
V. Sukhorukov; T. Baranich; T. Vinogradova; N. Klejmenova; V. Nevstrueva; A. Brydun; P. Shatalov; V. Glinkina; S. Artemyeva

G.P.65 **Mitochondrial disorders with novel mutations – A study from tertiary referral center in South India**
G. Narayanappa; C. Govindraju; P. Bindu; S. Kothari; A. Taly; K. Nahid; K. Thangaraj

G.P.66 **Phenotypic spectrum of patients with MERRF and mutations in the MTTK gene**
K. Claeys; J. Altmann; B. Büchner; A. Nadaj-Pakleza; D. Lehmann; L. Schöls; M. Deschauer; S. Jackson; J. Schäfer; R. Lautenschläger; A. Kuhn; J. Schulz; J. Weis; C. Kornblum; T. Klopstock

G.P.67 **Expanding the pathological phenotype in megaconial congenital muscular dystrophy**
G. Haliloglu; B. Talim; D. Aktas; M. Alikasifoglu; H. Topaloğlu

G.P.68 **Mitochondrial heat maps: A novel quantitative image analysis tool for assessing mitochondrial biogenesis in muscle biopsies**
R. Phadke; M. Ellis; A. Sigurta; S. Saeed; N. Hill; D. Chambers; M. Singer

G.P.69 **Structural muscle involvement in patients with chronic progressive external ophthalmoplegia assessed by quantitative MRI**
G. Hedermann; N. Løkken; J. Dahlqvist; C. Vissing; K. Knak; L. Andersen; C. Thomsen; J. Vissing

Myasthenia, myotonia, paramyotonia (G.P.70–84)
Facilitators: Marianne de Visser and Benedikt Schoser

G.P.70 **Myasthenia gravis and pregnancy: A brief review of 37 pregnancies**
R. Ducci; P. Lorenzoni; C. Kay; L. Werneck; R. Scola

G.P.71 **Serum BAFF levels in patients with myasthenia gravis**
S. Kang

G.P.72 **Familial MuSK antibody positive myasthenia gravis**
O. Ekmekci; A. Yuceyar; H. Karasoy

G.P.73 **Clinical characteristics of juvenile myasthenia gravis : A tertiary center experience**
H. Lee; H. Shin; H. Kang; S. Kim; J. Lee; H. Kim; S. Il Nam; Y. Lee

G.P.74 **Acetylcholine receptor and titin antibody in Chinese myasthenia gravis patients**
Y. Hong; H. Li; G. Skeie; N. Gilhus

G.P.75 **Very late-onset myasthenia gravis in Slovakia: Epidemiology and clinical characteristics**
P. Spalek; M. Fulova; I. Martinka; M. Spalekova; M. Soskova; I. Urminska

G.P.76 **A third case of congenital myasthenic syndrome caused by mutations in SCN4A**
N. Witting; U. Werlauf; M. Ballegaard; J. Vissing

G.P.77 **New SCN4A mutations – Unusual clinical phenotypes**
S. Sandell; J. Palmio; S. Penttilä; T. Suominen; B. Udd

G.P.78 **A Finnish mutation in SCN4A gene causes predominantly myalgic phenotype**
J. Palmio; S. Sandell; S. Penttilä; B. Udd

G.P.79 **Functional and pharmacological characterization of the new M1808I mutation in hNav1.4 found in a patient presenting with myotonia and myasthenia**
J. Roussel; P. Imbrici; M. Lorenzo; A. Farinato; G. Crescenzo; P. Bernasconi; R. Mantegazza; D. Conte Camerino; J. Desaphy

G.P.80 **Novel mutation in CACNA1S extends the phenotypic spectrum of periodic paralysis phenotypes: Electrodiagnostic and histopathological features**
S. Iyadurai; J. Roggenbuck; J. Kissel

G.P.81 **A novel mutation in a patient with a DOK7 congenital myasthenic syndrome**
T. Willis; R. Kulshrestha; C. Sewry; J. Palace; W. Liu; D. Beeson

G.P.82 **Schwartz–Jampel syndrome with learning difficulties and without osteochondrodysplasia**
S. Coppens; F. Ziereisen; G. Glibert; D. Valero-Hervás; S. Santillán; C. Vilain; N. Deconinck

G.P.83 **Congenital myasthenic syndromes in Israel: Genetic and clinical characterization**
S. Aharoni; M. Sadeh; L. Sagi; M. Rabie; M. Daana; Z. Argov; S. Edvardson; Y. Shapira; T. Dor; A. Bloch; A. Engel; Y. Nevo

G.P.84 **Fatigability during functional outcome measures in participants with myasthenic conditions**
V. Selby; G. Ramdharry; D. Balls; N. James; J. Hogrel; M. Hanna; F. Muntoni

FSHD (G.P.85–100)
Facilitators: Alexandra Belayew and Simon Hammans

G.P.85 **Perinatal complications in patients with congenital myotonic dystrophy**
M. Shichiji; K. Ishigaki; K. Ishiguro; T. Sato; T. Murakami; A. Muto; M. Osawa; S. Nagata

G.P.86 **Congenital myotonic dystrophy – Implications for cardiology screening**
A. Murphy; A. Porter; J. Bourke; V. Straub

G.P.87 **Clinical profile of congenital myotonic dystrophy**
C. Tian; J. Bange; I. Rybalsky; B. Wong

G.P.88 **Changes in clinical condition and causes of death of inpatients with myotonic dystrophy in Japan**
T. Saito; T. Takahashi; S. Kuru; M. Suzuki; T. Matsumura; H. Fujimura; S. Sakoda

G.P.89 **Myotonic dystrophy type 1 (DM1): Audiological and vestibular features found in Mexican patients**
M. Arenas-Sordo; B. Rivera-Mercado; D. Gutierrez-Tinajero; A. Martinez-Garcia Ramos; M. Trujillo-Bracamontes; O. Hernandez-Hernandez; J. Magaña-Aguirre

G.P.90 **Neuroglial miscommunication in the cerebellum of a mouse model of myotonic dystrophy**
L. Servais; G. Sicot; D. Dinca; G. Prigogine; A. Leroy; F. Medja; C. Chhuon; A. Huguet; A. Nicole; N. Gueriba; C. Guerrera; G. Cheron; G. Gourdon; M. Gomes-Pereira

G.P.91 **Why are FSHD muscles weak? A novel role for sarcomeric proteins**
S. Lassche; N. Voermans; A. Heerschap; M. Hopman; B. Kusters; S. van der Maarel; G. Stienen; C. Ottenheijm; B. van Engelen

G.P.92 **FSHD 1 and 2 testing – A clinical diagnostic service perspective**
D. Smith; R. Whittington; P. Lunt; T. Evangelista; M. Williams; H. Lochmüller

G.P.93 **Pain and quality of life in the UK FSHD patient registry**
T. Evangelista; L. Wood; M. Pohlschmidt; C. Longman; M. Roberts; D. Hilton-Jones; P. Lunt; T. Wills; R. Orrell; F. Norwood; M. Williams; D. Smith; J. Hudson; H. Lochmüller

G.P.94 **Computerized quantification of facial weakness in facioscapulohumeral muscular dystrophy**
K. Mul; S. van de Leemput; C. Horlings; C. Beurskens; S. Knuijt; G. Padberg; N. Voermans; F. Grootjen; B. van Engelen

G.P.95 **Disease modifying factors in facioscapulohumeral muscular dystrophy: Protocol of the FSHD-FOCUS study**
K. Mul; G. Horlings; R. Lemmers; N. Voermans; S. van der Maarel; B. van Engelen

G.P.96 **High intensity training in patients with facioscapulohumeral muscular dystrophy**
A. Buch; G. Andersen; K. Borup Heje Pedersen; J. Vissing

G.P.97 **The FSHD-related protein μ-crystallin controls metabolic and contractile properties in skeletal muscle**
D. Seko; T. Hisamatsu; R. Fujita; K. Yoshioka; Y. Urata; S. Goto; T. Li; A. Taimura; Y. Ono

G.P.98 **Baseline characteristics of the CINRG Infantile facioscapulohumeral muscular dystrophy cohort**
T. Duong; M. Eagle; J. Feng; J. Mah

G.P.99 **Resolaris, a potential therapeutic for rare myopathies with an immune component**
J. McKew; M. Ashlock; K. Ogilvie; L. Nangle; K. Taylor; K. Chiang; E. Mertsching; J. Chang; K. Moldt; C. Polizzi; Z. Xu; C. Lo; C. Lau; R. Adams; E. Chong

G.P.100 **Phenotype analysis of transgenic mice expressing mutant PABPN1**
T. Doki; S. Yamashita; T. Nishikami; Y. Nakajou; Y. Matsuo; N. Tawara; Y. Maeda; H. Hino; E. Uyama; K. Araki; Y. Ando

Muscle (G.P.101–111)
Facilitators: Helge Amthor and Mariz Vainzof

G.P.101 **Characterization of syndecan-4-dependent signalling events during myoblast proliferation**
A. Keller-Pinter; T. Kocsis; I. Ocsovszki; L. Dux

G.P.102 **Axial myopathy in a patient with a heterozygous MSTN mutation**
N. Voermans; B. Kusters; J. Rooy; R. Dieks; B. van Engelen; E. Kamsteeg; M. Schülke

G.P.103 **Control of transcription elongation is essential for cardiac and skeletal muscle development**
J. Bharj; M. Usyaloglu; D. Osborn; D. Zheng; Y. Jamshidi; F. Conti

G.P.104 **Investigation of the sources of osteopontin required for normal regeneration of injured muscle**
D.W. Wijesinghe; E. Mackie; C. Pagel

G.P.105 **The muscle environment affects the capacity of human myogenic progenitor cells to regenerate skeletal muscle and to reconstitute the satellite cell pool**
J. Meng; M. Bencze; R. Asfahani; F. Muntoni; J. Morgan

G.P.106 **The effect of antioxidants in muscle regeneration process**
A. Ishii; M. Yoshida; N. Ohkoshi; A. Tamaoka

G.P.107 **ACE-2494, a novel GDF ligand trap, increases muscle and bone mass upon systemic administration in mice**
R. Pearsall; D. Sako; J. Liu; M. Davies; K. Heveron; R. Castonguay; L. Krishnan; M. Troy; K. Liharska; R. Steeves; M. Cannell; M. Alimzhanov; A. Grinberg; R. Kumar

G.P.108 **ACE-083 increases muscle hypertrophy and strength in C57BL/6 mice**
R. Pearsall; J. Widrick; E. Cotton; D. Sako; J. Liu; M. Davies; K. Heveron; M. Maguire; R. Castonguay; L. Krishnan; M. Troy; K. Liharska; R. Steeves; J. Strand; T. Keefe; M. Cannell; M. Alimzhanov; A. Grinberg; R. Kumar

G.P.109 **Insulin-like growth factor binding protein 1 knockout increases free serum IGF-1 and enhances skeletal muscle mass and force in vivo**
W. Eilers; P. Crossey; K. Foster

G.P.110 **Does protease-activated receptor-2 (PAR_2) influence muscle development and regeneration?**
N. Taghavi Esfandouni; E. Mackie; C. Pagel

G.P.111 **Influence of mesenchymal stem cell on STAT signalling pathway in muscle satellite cell**
Y. Nakajou; Y. Maeda; Y. Yonemochi; T. Doki; N. Tawara; S. Yamashita; Y. Ando

15:45 **Afternoon refreshments, exhibition and posters –** Corn Exchange, café and mezzanine

16:00–17:30 **Guided poster discussion session 2: Parallel sessions (G.P.112–198)** – Corn Exchange

McArdle disease and related glycogenoses (G.P.112–122)
Facilitators: Claudio Bruno and Carmen Navarro

G.P.112 **Molecular, morphological and physiological studies in a mouse model of McArdle disease: Similarities to the human disease**
T. Krag; T. Pinos; T. Nielsen; A. Brull; A. Andreu; J. Vissing

G.P.113 **Clinical and molecular features of a large cohort of Italian McArdle patients**
D. Cassandrini; P. Tonin; L. Morandi; O. Musumeci; M. Filosto; G. Siciliano; E. Pegoraro; L. Santoro; R. Massa; T. Mongini; M. Sacchini; E. Bertini; G. Marrosu; M. Rigoldi; A. Burlina; A. Pini; S. Previtali; F. Santorelli; A. Toscano; C. Bruno

G.P.114 **Myophosphorylase (PYGM) mutations in Turkish patients with McArdle disease: A next generation sequencing study**
G. Inal Gültekin; B. Toptaş Hekimoğlu; Z. Görmez; H. Durmuş; H. Demirci; M. Sağıroğlu; Y. Parman; F. Deymeer; H. Yılmaz; S. Pençe; C. Kurt; E. Tan; S. Özdamar; U. Giger; O. Öztürk; P. Serdaroğlu-Oflazer

G.P.115 **Evaluation of maximum oxygen utilization in McArdle patients before and after exercise training**
O. Gelisin; H. Durmus; S. Yakal; E. Kasikcioglu; Y. Parman; F. Deymeer; P. Oflazer-Serdaroğlu

G.P.116 **McArdle disease: Clinical features, electrophysiological studies, muscle biopsy and molecular genetics**
P. Lorenzoni; R. Scola; C. Kay; R. Arndt; C. Silvado; L. Werneck

G.P.117 **Sodium valproate for McArdle disease (glycogen storage disease type V – GSDV)**
R. Scalco; C. Vissing; R. Godfrey; S. Chatfield; N. Løkken; K. Madsen; J. Pattni; Z. Michalak; C. Sewry; Z. Fox; G. McKenna; G. Samandouras; J. Holton; J. Howell; R. Haller; J. Vissing; R. Quinlivan

G.P.118 **Glycogen storage disease type XV: A case report**
A. Gardiner; R. Scalco; R. Pitceathly; J. Holton; M. Hanna; A. Schapira; H. Houlden; R. Lachmann; R. Quinlivan

G.P.119 **Clinical and paraclinical features of phosphoglucomutase type 1 deficiency: Biochemical clues for the diagnosis**
S. Souvannanorath; P. Laforêt; A. Bruneel; E. Morava; C. Sarret; O. Aumaitre; T. Dupre; T. Stojkovic

G.P.120 **Clinical, histopathological and molecular aspects of glycogen storage disease type VII: A review of the UK experience**
R. Godfrey; R. Scalco; C. Ellerton; R. Carruthers; S. Chatfield; K. Hansen; J. Michelson; J. Pattni; J. Holton; R. Quinlivan

G.P.121 **A novel neuromuscular form of glycogen storage disease type IV characterized by spinal stiffness, arthrogrypotic features and rare polyglucosan bodies in muscle biopsy**
E. Malfatti; C. Barnerias; C. Hedberg-Oldfors; C. Gitiaux; V. Allamand; R. Carlier; A. Oldfors; S. Quijano-Roy; N. Romero

G.P.122 **Unusual demyelinating pathology in a case of adult polyglucosan body disease**
R. Phadke; E. Murphy; O. Cicarrelli; M. Koltzenburg; R. Kirk; M. Lunn

SMA non-5q (G.P.123–136)
Facilitators: Helen Roper and David Hilton-Jones

G.P.123 **Distal spinal muscular atrophy and ataxia with cerebellar atrophy in two unrelated patients; a new phenotypic variant of HRD and recessive KCS syndrome related to TBCE**
E. Bertini; A. Sferra; T. Rizza; G. Tasca; A. D'Amico; G. Zanni; S. Barresi; D. Diodato; E. Piermarini; D. Martinelli; C. Dionisi-Vici; M. Niceta; B. Dallapiccola; M. Tartaglia; C. Compagnucci

G.P.124 **Spinal Muscular Atrophy-Lower Extremity Dominant (SMA-LED) with bilateral perisylvian polymicrogyria and infantile epileptic encephalopathy due a novel DYNC1H1 mutation**
J. Singh; M. Illingworth; A. Whitney; D. Konn; N. Foulds; D. Allen; M. Uglow

G.P.125 **Hereditary spastic paraplegia and peroxisome biogenesis disorders: Case report of a patient with mutations in PEX10 gene**
A. Nascimento; C. Ortez; A. Sariego; E. Gerotina; J. Armstrong; C. Sierra; R. Artuch; C. Jou; C. Jimenez-Mallebrera; J. Colomer

G.P.126 **A new family with autosomal recessive spastic ataxia of Charlevoix-Saguenay. ARSACS**
J. Palmio; S. Penttilä; J. Moilanen; M. Kärppä; B. Udd

G.P.127 **Elucidation of the mechanism of disease in DYNC1H1-associated motor neuron disease and potential therapeutic targets**
M. Yang; C. Kearns; E. McGonagle; B. Appel

G.P.128 **Occurrence of CHCHD10 mutations in Finnish patients with motor neuron disorder**
S. Penttilä; M. Jokela; A. Saukkonen; J. Toivanen; B. Udd

G.P.129 **TRPV4 mutation causing dominantly inherited scapuloperoneal spinal muscle atrophy**
P. Sanaker; L. Bindoff

G.P.130 **Atypical presentation of SMARD1**
R. Kulshrestha; N. Kiely; T. Willis

G.P.131 **Next generation sequencing identifies a novel ATP7A mutation in two brothers with distal hereditary motor neuropathy and autonomic dysfunction**
C. Scotton; E. Italyankina; M. Storbeck; K. Vezyroglou; R. Heller; M. Neri; F. Di Raimo; A. Mauro; V. Tugnoli; V. Timmerman; B. Wirth; D. De Grandis; F. Gualandi; A. Ferlini

G.P.132 **Mutations in ASAH1 may cause spinal muscular atrophy**
A. Behin; I. Nelson; G. Bonne; N. Romero; R. Froissart

G.P.133 **Modeling and pathophysiological analysis of the ubiquilin-2-linked amyotrophic lateral sclerosis (ALS) with AAV vectors**
C. Bos; M. Biferi; M. Cohen-Tannoudji; M. Roda; M. Barkats

G.P.134 **High intensity training in patients with spinal and bulbar muscular atrophy**
K. Heje; G. Andersen; J. Vissing

G.P.135 **Effects of a program based on home exercise on muscle force and functional independence in patients with amyotrophic lateral sclerosis: A 2-year follow-up study**
M. Piemonte; C. Ramirez; M. Voos; T. Oliveira; C. Miranda; H. Silva; D. Callegaro

G.P.136 **Survey of clinicians experience and attitudes regarding spinal muscular atrophy with respiratory distress syndrome type 1 (SMARD1) in the UK**
S. Shah; T. Jerrom; J. Fraser; A. Majumdar

DMD functional scales (G.P.137–154)
Facilitators: Michelle Eagle and Laurent Servais

G.P.137 **Arm function of boys with Duchenne muscular dystrophy explored by surface electromyography and muscle force measurements**
M. Janssen; J. Harlaar; I. de Groot

G.P.138 **Determinants to predict upper extremity function in boys and men with DMD**
M. Janssen; J. Hendriks; A. Geurts; I. de Groot

G.P.139 **Development of a patient-reported outcome measure for arm and hand function in Duchenne muscular dystrophy (UL-PROM DMD)**
K. Klingels; A. Mayhew; E. Mazzone; M. Main; V. Decostre; M. van den Hauwe; M. Eagle; T. Duong; I. de Groot; V. Ricotti; U. Werlauff; G. Campion; E. Vroom; E. Mercuri; N. Goemans

G.P.140 **Exploring the relationships between grip and pinch strength and functional scales**
J. Hogrel; ULENAP Study Group

G.P.141 **Upper limb function in Duchenne muscular dystrophy – A natural history investigating upper limb performance relative to ambulatory stage**
J. Goubran; K. Carroll; M. Ryan

G.P.142 **Reachable workspace and performance of upper limb (PUL) in Duchenne muscular dystrophy**
C. McDonald; J. Han; E. de Bie; A. Nicorici; R. Abresch; C. Anthonisen; R. Bajcsy

G.P.143 **A newly developed six-minute walking test for Duchenne muscular dystrophy**
H. Genno; H. Nishizawa; N. Shiba; A. Nakamura

G.P.144 **Reference values for the three-minute walk test, North Star ambulatory assessment and timed tests in typically developing boys aged 2.5–5 years**
K. Klingels; L. Van Verdegem; M. vanden Hauwe; G. Buyse; N. Goemans

G.P.145 **Assessment of early motor development in young boys with Duchenne muscular dystrophy**
K. Klingels; M. van den Hauwe; F. Smeets; M. Schraeyen; G. Buyse; N. Goemans

G.P.146 **A latent factor-based composite timed motor performance index representing mobility is sensitive to stage of disease and 1-year progression in Duchenne muscular dystrophy**
E. Henricson; R. Abresch; E. Goude; A. Nicorici; E. deBie; C. McDonald

G.P.147 **Outcome measures for Duchenne muscular dystrophy from ambulant to non-ambulant: Implications for clinical trials**
V. Ricotti; M. Eagle; J. Butler; V. Decostre; R. Deborah; A. Moraux; K. Anthony; V. Sleby; M. Guglieri; M. van der Holst; M. Jansen; J. Morgan; I. de Groot; E. Niks; J. Verschuuren; L. Servais; J. Hogrel; T. Voit; V. Straub; F. Muntoni

G.P.148 **Validity of ACTIVE for use across ambulatory abilities in Duchenne muscular dystrophy**
L. Lowes; L. Alfano; K. Berry; K. Flanigan; J. Mendell

G.P.149 **Progression of timed performance and compensatory movements on locomotion tasks (10-m walking and climbing up and down steps) in children with Duchenne muscular dystrophy**
M. Voos; J. Martini; F. Caromano

G.P.150 **The relevance of timed movements on functional assessment in Duchenne muscular dystrophy**
M. Voos; J. Martini; M. Simões; M. Hukuda; F. Favero; A. Oliveira; F. Caromano

G.P.151 **Responsiveness of the Functional Evaluation Scale for patients with Duchenne muscular dystrophy – Domain 1: Sitting and rising from the chair**
M. Voos; M. Hukuda; E. de Carvalho; F. Caromano

G.P.152 **Functional and gait assessment in children with Duchenne muscular dystrophy: Quantitative and functional evaluation**
G. Vasco; M. Petrarca; A. D'Amico; G. Colia; M. Catteruccia; S. Gazzellini; S. Carniel; A. Pisano; E. Castelli; E. Bertini

G.P.153 **Responsiveness of the Functional Evaluation Scale for patients with Duchenne muscular dystrophy – Domain 2: Gait**
E. de Carvalho; M. Hukuda; M. Voos; F. Caromano

G.P.154 **Longitudinal functional measures in Becker muscular dystrophy: Implications for clinical trials and Duchenne exon skipping outcomes**
L. Bello; P. Campadello; A. Barp; C. Semplicini; G. Soraru; C. Angelini; E. Pegoraro

LGMD (G.P.155–173)
Facilitators: Kate Bushby and Isabelle Richard

G.P.155 **Preparation of a disease specific functional measure suitable for trials in dysferlinopathy**
A. Mayhew; M. James; M. Eagle; C. Bladen; R. Muni Lofra; K. Bettinson; H. Hilsden; E. Harris; K. Bushby; L. Rufibach; The Jain COS Consortium

G.P.156 **The importance of dosage analysis in dysferlinopathy**
J. Hudson; E. Graham; R. Charlton; M. Guglieri; K. Bushby; V. Straub; R. Barresi

G.P.157 **High expression and therapeutic efficacy of systemic delivery of a dual AAV strategy in a murine model for dysferlin deficiency**
M. Pryadkina; C. Roudaut; K. Charton; W. Lostal; M. Hirsch; N. Bourg; I. Richard;

G.P.158 **Description of muscle strength in a Brazilian sample of limb-girdle muscular dystrophy 2A (calpainopathy)**
M. Voos; J. Marim; I. Anequini; F. Favero; A. Carmo; A. Oliveira; F. Caromano

G.P.159 **Complex relationship between calpain 3 and titin**
K. Charton; J. Sarparanta; A. Milic; A. Vihola; P. Jonson; B. Udd; I. Richard

G.P.160 **CAPN3 mutation c. 643-663del21 identified in 52 Norwegian patients belonging to 24 families is associated with dominant calpainopathy**
M. Van Ghelue; C. Wahl; K. Arntzen; S. Loseth; L. Bindoff; L. Sveberg; O. Rosby; T. Popperud; M. Rasmussen; H. Halvorsen; S. Lindal; C. Jonsrud; K. Orstavik

G.P.161 **Muscle MRI in sarcoglycanopathies**
G. Tasca; M. Monforte; G. Brisca; A. D'Amico; N. Al Shaik; A. Berardinelli; N. Løkken; L. Maggi; L. Morandi; N. Voermans; F. Munell; A. Sanchez; J. Dastgir; E. Bertini; C. Bruno; MYO-MRI COST Study Group

G.P.162 **Frequency and natural history of sarcoglycanopathies in Paris neuromuscular centers**
R. Guimarães-Costa; A. Behin; T. Stojkovic; S. Leonard-Louis; I. Desguerre; C. Barnerias; M. Mayer; A. Isapof; B. Estournet; F. Leturcq; C. Semplicini; B. Eymard; P. Laforêt

G.P.163 **Probable high prevalence of limb-girdle muscular dystrophy type 2D in the aboriginal tribes settled in southern Taiwan**
W. Liang; P. Chou; T. Kan; Y. Su; Y. Hayashi; I. Nishino; Y. Jong

G.P.164 **Natural history study of mouse models for limb girdle muscular dystrophy, types 2D and 2F**
M. van Putten; C. Tanganyika-de Winter; J. van der Meulen; L. van Vliet; J. Plomp; A. Aartsma-Rus

G.P.165 **Discovery of biomarkers for LGMD2I.**
R. Heredia; Y. Hathout; S. Moore; M. Katherine; S. Cirak

G.P.166 **Muscle strength relative to cross-sectional area in hypertrophic calf muscles of patients affected by limb girdle type 2I and Becker muscular dystrophies**
N. Løkken; G. Hedermann; C. Thomsen; J. Vissing

G.P.167 **Prognostic features of LGMD presenting as symptomatic or paucisymptomatic hyperCKemia**
P. Marti; N. Muelas; O. Jaka; F. Mayordomo; I. Azorin; A. Lopez de Munain; P. Gallano; J. Vilchez

G.P.168 **Clinical data and MRI findings in two brothers with limb girdle muscular dystrophy due to LAMA2 mutations**
E. Harris; A. Topf; M. McEntagart; C. Sewry; V. Straub

G.P.169 **Late-onset limb-girdle muscular dystrophy caused by GMPPB-mutation**
H. Balcin; J. Palmio; I. Nennesmo; G. Solders; B. Udd

G.P.170 **Novel histopathological features in a patient with a novel mutation in DNAJB6 gene (LGMD 1D locus)**
S. Iyadurai; J. Roggenbuck; M. Freimer

G.P.171 **LGMD1F: A morphological study**
B. San Millan; S. Ortolano; S. Teijeira; J. Gamez; A. Andreu; R. Marti; C. Navarro

G.P.172 **Molecular interaction of caveolin-3 and nNOS: Implication in the pathogenesis of limb-girdle muscular dystrophy 1C**
Y. Ohswa; Y. Fukai; S. Nishimatsu; M. Fujino; Y. Sunada

G.P.173 **A novel mutation in DNAJB6 causes LGMD1D in two French families**
P. Jonson; A. Evilä; T. Stojkovic; F. Chapon; H. Luque; P. Hackman; B. Udd

IBM (G.P.174–184)
Facilitators: Yves Allenbach and Jan De Bleecker

G.P.174 **Myofiber HLA-DR expression is a distinctive biomarker for overlap myositis and inclusion body myositis**
J. Aouizerate; Y. Baba Amer; G. Bazzez; R. Gherardi; M. Rigoloet; F. Authier

G.P.175 **Cardiac involvement of sporadic inclusion body myositis detected by cardiac magnetic resonance imaging**
A. Rosenbohm; D. Buckert; J. Kassubek; W. Rottbauer; A. Ludolph; P. Bernhardt

G.P.176 **Agreeing best practice guidelines for inclusion body myositis**
M. Rose; K. Jones; IBM Study Group UK

G.P.177 **Experience with abatacept treatment in sporadic inclusion body myositis**
C. Lindberg; A. Ewaldsson; U. Edofsson; E. Hammarén

G.P.178 **Study design of a prospective natural history study in sporadic inclusion body myositis (sIBM)**
P. Houston; L. Lowes; S. Greenberg; P. Machado; D. Papanicolaou; A. Genge

G.P.179 **Rimmed vacuoles are positive for RIPK1 and RIPK3**
T. Kurashige; H. Morino; Y. Nagano; H. Maruyama; M. Matsumoto

G.P.180 **Resource utilization in a US-based sample of patients with sporadic inclusion body myositis (sIBM)**
C. DeMuro; V. Williams; R. Goldberg; L. Lowes; M. Price; G. Capkun; V. Barghout; B. Tseng

G.P.181 **Mitochondrial pathology in inclusion body myositis**
U. Lindgren; S. Roos; C. Hedberg Oldfors; A. Moslemi; C. Lindberg; A. Oldfors

G.P.182 **A single center analysis of the clinicopathological findings of anti-cytosolic 5′-nucleotidase 1a antibody-positive sporadic inclusion body myositis**
N. Tawara; S. Yamashita; T. Nishikami; Y. Nakajou; T. Doki; Y. Matsuo; Y. Yonemochi; Y. Maeda; Y. Ando

G.P.183 **Psychometric evaluation of the sporadic inclusion body myositis (sIBM) physical functioning assessment (sIFA)**
V. Williams; T. Coles; C. DeMuro; S. Lewis; N. Williams; S. Yarr; V. Barghout; L. Lowes; L. Alfano; B. Goldberg; A. Gnanasakthy; G. Capkun; B. Tseng

G.P.184 **Sporadic inclusion body myositis misdiagnosed as granulomatous myositis**
T. Mozaffar; M. Lavian; N. Goyal

Miscellaneous (G.P.185–198)
Facilitators: Mayana Zatz and Matt Parton

G.P.185 **Assessment of a panel of antibodies to myosin heavy chains applied to human skeletal muscle biopsies**
L. Feng; D. Chambers; N. Bhardwaj; R. Phadke; F. Muntoni; C. Sewry

G.P.186 **Establishing normative data for the 60 and 100-meter run test in healthy male controls**
L. Lowes; L. Alfano; K. Berry; M. Leggett

G.P.187 **Clinical assessment of respiratory dysfunction in children with neuromuscular diseases**
C. Bozdemir Özel; N. Bulut; H. Alkan; I. Öncel; O. Yılmaz; A. Karaduman

G.P.188 **Comparison of ACTIVE-mini to the Vicon motion camera system in measuring infant movements**
L. Lowes; M. McNally; L. Alfano; K. Berry; J. Mendell; J. Heathcock

G.P.189 **Ischemic inflammatory myopathy with microangiopathy in Fabry disease**
K. Benistan; R. Carlier; L. Echevarria; S. Quijano-Roy; D. Germain; F. Authier

G.P.190 **The feasibility of rare neuromuscular disease trials: Presenting the "clinical research unit" as a model framework**
A. Genge; N. Campbell

G.P.191 **Neuromuscular patients: Complaints are unrelated to disease**
J. Forbes; C. Riolfi; E. Macedo; T. Genesini; A. Mouzat; D. Rüdiger; G. Achôa; H. Andrade; L. Lise; L. Fachinetto; M. Vianna; R. Pavanello; M. Zatz

G.P.192 **The floppy infants: Clinical experience in tertiary centers for the past 10 years**
H. Lee; Y. Lee; H. Kang; J. Lee; H. Kim; S. Lee; M. Park; R. Namkung; J. Lee; H. Chung

G.P.193 **Clinical psychology provision in the neuromuscular team**
Y. Easthope-Mowatt; R. Kulshrestha; T. Willis

G.P.194 **Evaluation of patient experience of transition in the neuromuscular clinic**
T. Willis; I. Mahabeer; Z. Alhaswani

G.P.195 **Unilateral tensor fascia lata muscle mass as a presentation of lumbosacral radiculopathy**
P. Soltanzadeh; B. Pierce; S. Lietman; H. Ilaslan

G.P.196 **Quantifiable diagnosis of neuromuscular diseases through network analysis**
E. Rivas; A. Sáez; A. Montero-Sánchez; C. Paradas; B. Acha; A. Pascual; C. Serrano; L. Escudero

G.P.197 **Triple trouble – DMD, autism, epilepsy**
L. Mrazova; Z. Jurikova; P. Danhofer; J. Pejcochova; P. Vondracek; J. Zamecnik; T. Honzik; H. Oslejskova

G.P.198 **Life as a teenage girl with NMD in Denmark is good – And difficult**
A. Hoejberg

17:30–19:30 **Symposium 1; Sarepta Therapeutics** – Concert Hall, including refreshments in Corn Exchange

Friday 2 October 2015

08:30 **Conference desk opens** – Brighton Dome foyer

07:30–08:45 ***Neuromuscular Disorders* Editorial Board Meeting** – William IV Room, Brighton Pavilion

09:00–10:30 **Immune-mediated peripheral nerve, neuromuscular junction and muscle disorders;** Invited lectures (I.I.5–7) – Concert Hall
Chairpersons: Peter Van den Bergh and George Padberg

I.I.5 **Autoimmune neuropathies: Pathogenesis and future perspectives**
H. Willison

I.I.6 **Myasthenia: Novel antigens and therapies**
J. Verschuuren

I.I.7 **Contribution of myopathological analysis to myositis classification in a multidisciplinary approach**
W. Stenzel

10:30–11:00 **Morning refreshments, exhibition and posters** – Corn Exchange, café and mezzanine

11:00–11:30 **Immune-mediated peripheral nerve, neuromuscular junction and muscle disorders;** Invited lecture (I.I.8) – Concert Hall
Chairpersons: Caroline Sewry and Thomas Voit

I.I.8 **Genomic insights into inflammatory myopathies**
S. Greenberg

11:30–13:00 **Immune-mediated peripheral nerve, neuromuscular junction and muscle disorders;** Oral Presentations (G.O.7–12) – Concert Hall
Chairpersons: Caroline Sewry and Thomas Voit

G.O.7 **Choosing the "best" animal model for pre-clinical trials of autoimmune myasthenia gravis**
G. Shelton

G.O.8 **Cancer and necrotizing immune myopathy: High incidence in anti-HMGCR positive and seronegative patients but not in anti-SRP positive patients**
Y. Allenbach; J. Kaeren; A. Bouvier; N. Champtiaux; N. Shoindre; K. Mariampillai; A. Rigolet; L. Musset; P. Laforêt; B. Eymard; T. Stojkovic; A. Behin; B. Hervier; O. Benveniste

G.O.9 **Integrins are required for synaptic transmission and development of the neuromuscular junction**
J. Ross; R. Webster; T. Lechertier; L. Reynolds; F. Muntoni; D. Beeson; K. Hodivala-Dilke; F. Conti

G.O.10 **Exome sequencing identifies novel truncating TTN mutations with Emery–Dreifuss like muscular dystrophy and secondary calpain3 deficiency without cardiac abnormality**
R. de Cid; R. Ben Yaou; C. Roudaut; S. Baulande; F. Leturcq; G. Bonne; B. Udd; N. Romero; K. Charton; E. Malfatti; I. Nelson; B. Eymard; I. Richard

G.O.11 **RNA profiling discloses a link between circadian genes and muscle damage in Duchenne muscular dystrophy**
C. Scotton; A. Armaroli; H. Osman; M. Falzarano; R. Capogrosso; A. Cozzoli; G. Camerino; A. De Luca; A. Ferlini

G.O.12 **Muscular dystrophy (*mdx*) mice with enhanced voluntary exercise: Mining genetic modifiers**
A. Messineo; R. Chidambaram; E. McNamara; G. Manship; A. Wallace; C. Murray; M. Mehta; Q. Nguyen; R. Ram; N. Laing; G. Morahan; K. Nowak

13:00–15:00 **Lunch** – Corn Exchange, café and mezzanine

13:30–15:00 **Symposium 2; PTC Therapeutics** – Concert Hall

15.00–16:30 **Guided poster discussion session 3: Parallel sessions (G.P.199–308)** – Corn Exchange

Inflammatory muscle diseases (G.P.199–212)
Facilitators: Hans Goebel and François-Jerôme Authier

G.P.199 **Roles of hypoxia and innate immune mechanisms in juvenile and adult dermatomyositis**
C. Preuße; Y. Allenbach; H. Goebel; D. Pehl; J. Radke; U. Schneider; M. Vorgerd; A.v.Moers; B. Schoser; U. Schara; W. Stenzel

G.P.200 **Role of hypoxia in innate immunity activation in dermatomyositis**
E. Gallardo; X. Suarez-Calvet; N. De Luna; R. Rojas-Garcia; J. Diaz-Manera; I. Illa

G.P.201 **Phenotype-antibody correlation in dermatomyositis (DM)**
N. Chahin

G.P.202 **Vasculopathy as a major marker of severity in juvenile dermatomyositis**
C. Gitiaux; M. De Antonio; J. Aouizerate; R. Gherardi; T. Guilbert; C. Barnerias; C. Bodemer; K. Brochard-Payet; P. Quartier; L. Musset; B. Chazaud; I. Desguerre; B. Bader-Meunier

G.P.203 **How do tissue infiltrating B cells correlate with other inflammatory features in muscle tissue from patients with JDM and their clinical parameters?**
E. Sag; S. Yasin; K. Arnold; J. Holton; T. Jacques; L. Wedderburn

G.P.204 **Inflammatory milieu of muscle biopsies and clinical features in juvenile dermatomyositis**
E. Sag; S. Ozen; G. Kale; H. Topaloğlu; B. Talim

G.P.205 **Pattern of perifascicular involvement differentiates the anti-Jo1 inflammatory myopathy (IM) from dermatomyositis (DM)**
N. Chahin

G.P.206 **A case of chronic sarcoid myopathy with IBM like distal muscle involvement**
H. Karasoy; M. Eroglu; F. Gokcay; M. Argin; O. Ekmekci; A. Yuceyar

G.P.207 **Atypical presentation of childhood-onset inflammatory myositis. report of three cases**
A. van der Kooi; B. Jaeger; E. Aronica; M. de Visser

G.P.208 **Early onset autoimmune necrotizing myopathy associated with anti-HMGCR antibodies: An unmissable diagnosis**
A. Paipa Merchan; C. Jou; S. Kapetanovic; V. Velez; M. Fernández Cuesta; F. García-Bragado; C. Ortez; N. Vidal; C. Jimenez-Mallebrera; J. Colomer; A. Nascimento; M. Olivé

G.P.209 **Plasma IP-10 level distinguishes inflammatory myopathy**
A. Uruha; S. Noguchi; W. Sato; H. Nishimura; S. Mitsuhashi; T. Yamamura; I. Nishino

G.P.210 **Myopathy with anti signal recognition particle antibodies: Clinical and HLA associations in Malaysian patients**
T. Ambang; H. Shanmugam; J. Tan; C. Tan; T. Cheah; S. Sargunan; N. Shahrizaila; K. Wong; K. Goh

G.P.211 **Effects of auto-antibodies anti-signal recognition particle (SRP) and anti -hydroxymethylglutaryl-CoA reductase (HMGCR) on muscle cells**
L. Arouche-Delaperche; G. Butler-Browne; O. Benveniste

G.P.212 **Fasciitis frequently accompanies myopathy in acute critical illness muscle wasting: Evidence from qualitative ultrasound and muscle biopsy analysis**
R. Phadke; Z. Puthucheary; J. Rawal; M. McPhail; P. Sidhu; A. Rowlerson; J. Moxham; S. Harridge; N. Hart; H. Montgomery

DMD biomarkers; (G.P.213–229)
Facilitators: Annemieke Aartsma-Rus and Alessandra Ferlini

G.P.213 **Glutathione imbalance in blood of patients with Duchenne muscular dystrophy**
S. Petrillo; F. Piemonte; M. Catteruccia; E. Bertini; A. D'Amico

G.P.214 **Brain metabolite concentrations in Duchenne muscular dystrophy are unaltered compared to controls**
N. Doorenweerd; E. Niks; C. Straathof; A. Webb; J. Hendriksen; J. Verschuuren; M. van Buchem; H. Kan

G.P.215 **Mild course in atypical Duchenne muscular dystrophy patient is not caused by utrophin overexpression**
M. Vainzof; L. Feitosa; M. Canovas; R. Pavanello; M. Zatz

G.P.216 **Biomarker development to support the clinical development of utrophin modulators for Duchenne muscular dystrophy therapy**
J. Tinsley; N. Janghra; J. Morgan; C. Sewry; F. Muntoni; D. Elsey; F. Wilson; K. Davies

G.P.217 **Identification of novel therapy-responsive protein biomarkers for Duchenne muscular dystrophy by aptamer-based serum proteomics**
A. Coenen-Stass; G. McClorey; R. Manzano; C. Betts; A. Blain; A. Saleh; M. Gait; H. Lochmüller; L. Gold; M. Wood; T. Roberts

G.P.218 **Reversible immortalization allows human artificial chromosome-mediated gene correction of human dystrophic muscle progenitor cells**
S. Benedetti; H. Hoshiya; M. Ragazzi; N. Uno; Y. Kazuki; G. Ferrari; R. Tonlorenzi; A. Lombardo; V. Mouly; L. Naldini; G. Messina; M. Oshimura; G. Cossu; F. Tedesco

G.P.219 **miRNAs as serum biomarkers for Duchenne muscular dystrophy: Correlation analysis in a multicentre study between miRNAs levels and clinical status of DMD patients**
A. D'Amico; J. Martone; S. Previtali; G. Baranello; G. D'Angelo; A. Berardinelli; S. Messina; G. Vita; M. Pane; L. Morandi; E. Mercuri; M. Catteruccia; E. Bertini; I. Bozzoni

G.P.220 **Inflammatory- and NaCl-induced expression of NFAT5 in muscle cells point to common stress response mechanisms**
S. Herbelet; K. Schmidt; J. Zschuntzsch; E. Nys; L. Weynants; B. De Paepe; O. De Wever; J. De Bleecker; J. Schmidt

G.P.221 **Tissue- and case-specific retention of intron 40 in mature dystrophin mRNA**
A. Nishida; M. Minegishi; A. Takeuchi; T. Niba; H. Awano; T. Lee; K. Iijima; Y. Takeshima; M. Matsuo

G.P.222 **Nonsense mutation dystrophinopathy: How mutation-specific treatments changed our clinical practice?**
D. Ardicli; B. Konuskan; G. Haliloglu; M. Alikasifoglu; H. Topaloğlu

G.P.223 **Gene expression profiling of blood in patients with Duchenne muscular dystrophy**
B. Wong; D. Liu; S. Hu; P. Morehart; B. Stamova; B. Ander; F. Sharp

G.P.224 **microRNA and mRNA expression networks in Duchenne muscular dystrophy**
B. Wong; D. Liu; B. Stamova; S. Hu; P. Morehart; B. Ander; G. Jickling; X. Zhan; F. Sharp

G.P.225 **Breakpoint spanning PCR of the DMD gene to resolve discrepancy between MLPA and targeted aCGH results to confirm predicted effect on reading frame**
E. Ulm; M. Hegde; B. Ramesh Reddy Nallamilli; J. Alexander; B. Wong

G.P.226 **Quantifying dystrophin in cell culture: A method to accelerate preclinical assessment of DMD treatments**
E. Ruiz-Del-Yerro; I. García; V. Arechavala-Gomeza

G.P.227 **Genetic polymorphisms modify intramuscular fat infiltration in Duchenne muscular dystrophy**
A. Barnard; S. Romero de Mello Sa; R. Willcocks; C. Senesac; R. Finkel; S. Forbes; H. Sweeney; G. Tennekoon; W. Triplett; D. Lott; D. Wang; B. Byrne; D. Hammers; J. Pham; W. Rooney; E. Finanger; G. Walter; K. Vandenborne; B. Russman

G.P.228 **Micro RNA profile associated with the dystrophin level in Becker muscular dystrophy**
I. Zaharieva; S. Cirak; K. Anthony; L. Feng; G. Tasca; A. Ferlini; J. Morgan; F. Muntoni

G.P.229 **Mutation spectrum of the dystrophin gene in 507 Korean Duchenne/Becker muscular dystrophy patients**
H. Ryu; A. Cho; M. Seong; S. Park; J. Lee; B. Lim; K. Kim; Y. Hwang; J. Chae

Myofibrillar myopathies (G.P.230–242)
Facilitators: Montse Olivé and Kristl Claeys

G.P.230 **Phenotypic variability of Pro209Leu BAG3 mutations: Cardiomyopathy, demyelinating neuropathy and a Long QT syndrome**
A. Kostera-Pruszczyk; R. Ploski; A. Potulska-Chromik; E. Sadurska; P. Pruszczyk; J. Karolczak; M. Redowicz; A. Kaminska

G.P.231 **A novel phenotype associated with STIM1 gene: A case report of a patient with a painful myopathy**
M. Neri; E. Harris; C. Scotton; A. Topf; M. Fang; J. Wang; R. Barresi; H. Lochmüller; V. Straub; A. Ferlini

G.P.232 **Myofibrillar myopathies – An expanding spectrum of disorders**
G. Narayanappa; S. Rashmi; A. Nalini; A. Taly

G.P.233 **Mutated HSPB8 causes both neurogenic and myopathic disease with muscle proteinopathy**
M. Lindfors; R. Ghaoui; S. Penttila; J. Palmio; M. Needham; K. North; N. Clarke; C. Sue; P. Jonson; A. Vihola; B. Udd

G.P.234 **Investigating the pathobiology of myofibrillar myopathies and potential therapies using zebrafish**
A. Ruparelia; V. Oorschot; G. Ramm; R. Bryson-Richardson

G.P.235 **Mitochondrial dysfunction: A key player in myofibrillar myopathy?**
A. Vincent; M. Rocha; K. Rygiel; J. Grady; G. Campbell; R. Barresi; R. Taylor; D. Turnbull

G.P.236 **Impact of environmental stress and new models for pathophysiological and therapeutic studies of desminopathies**
F. Delort; B. Segard; P. Joanne; O. Agbulut; P. Vicart; S. Batonnet-Pichon

G.P.237 **Lethal multiple pterygium syndrome associated with mutations in the type 1 ryanodine receptor (RYR1)**
A. Kariminejad; S. Ghaderi-Sohi; H. Hossein-Nejad Nedai; V. Varasteh; H. Tajsharghi

G.P.238 **RYR1-related exertional rhabdomyolysis: Expanding spectrum and diagnostic challenges**
A. Gardiner; R. Scalco; M. Parton; M. Hanna; R. Pitceathly; E. Zanoteli; E. Murphy; S. Treves; H. Houlden; J. Wilmshurst; V. Straub; D. Hilton-Jones; N. Voermans; A. Manzur; P. Oflazer; U. Reed; R. Lachmann; R. Quinlivan; H. Jungbluth; On behalf of RM study group

G.P.239 **Zebrafish models of inherited skeletal muscle disorders**
J. Patrick; N. Wali; I. Sealy; J. Collins; E. Busch-Nentwich; D. Stemple

G.P.240 **Clinical. pathology and imaging heterogeneity in autosomal recessive RYR1-related myopathy**
G. Tasca; F. Fattori; D. Cassandrini; M. Catteruccia; M. Verardo; G. Vasco; M. Monforte; E. Ricci; E. Bertini; A. D'Amico

G.P.241 **Literature synthesis of RyR1 and its interacting proteins**
J. Witherspoon; M. Jain; K. Meilleur

G.P.242 **Neuromuscular conditions associated with malignant hyperthermia in paediatric patients: A 25-year retrospective study**
A. Bamaga; S. Riazi; K. Amburgey; W. Halliday; A. Guerguerian; J. Dowling; G. Yoon

Drug therapies for DMD (G.P.243–260)
Facilitators: Dominic Wells and Nathalie Goemans

G.P.243 **First drug registry in Duchenne muscular dystrophy (DMD) to assess Translarna (Ataluren) use, safety, and effectiveness in routine clinical practice**
K. Bushby; A. Reha; V. Northcutt; X. Luo; T. Ong; S. Park; R. Spiegel; Translarna Patient Registry

G.P.244 **Development of Rimeporide. a sodium-hydrogen exchanger (NHE-1) inhibitor, for patients with Duchenne muscular dystrophy**
F. Porte-Thomé; K. Nagaraju; Q. Yu; K. Tatem; G. Bkaily; W. Scholz; A. Slade; N. Bot; C. Kant

G.P.245 **Second generation utrophin modulator for the therapy of Duchenne muscular dystrophy**
S. Guiraud; S. Squire; B. Edwards; H. Chen; D. Burns; N. Shah; A. Babbs; S. Davies; G. Wynne; A. Russell; D. Elsey; F. Wilson; J. Tinsley; K. Davies

G.P.246 **Utrophin modulators significantly improve muscular dystrophy in the *mdx* diaphragm**
S. Guiraud; H. Chen; S. Squire; B. Edwards; D. Burns; N. Shah; S. Davies; G. Wynne; A. Russell; D. Elsey; F. Wilson; J. Tinsley; K. Davies

G.P.247 **Discovery of small molecule utrophin modulators for the therapy of Duchenne muscular dystrophy (DMD)**
N. Araujo; A. Vuorinen; R. Fairclough; S. Guiraud; J. Donald; C. Cairnduff; D. Hewings; F. Martinez; K. Csatayova; N. Willis; S. Squire; A. Babbs; B. Edwards; N. Shah; J. Tinsley; F. Wilson; S. Davies; G. Wynne; K. Davies; A. Russell

G.P.248 **Therapeutic potential of adiponectin and adipoRon in Duchenne muscular dystrophy**
S. Lecompte; M. Abou-Samra; R. Boursereau; L. Noel; S. Brichard

G.P.249 **Utrophin modulators to treat Duchenne muscular dystrophy (DMD): Results from a Phase 1b Clinical Trial of SMT C1100**
J. Tinsley; F. Muntoni; S. Spinty; H. Roper; I. Hughes; R. Ricotti; B. Tejura; G. Layton; K. Davies

G.P.250 **Small molecule compounds that promote exon skipping in the DMD gene**
N. Naryshkin; A. Dakka; V. Gabbeta; J. Pichardo; P. Vazirani; G. Ryan; M. Woll; A. Turpoff; N. Zhang; S. Zhang; G. Karp; E. Welch

G.P.251 **Exon 53 skipping of the dystrophin gene in patients with Duchenne muscular dystrophy by systemic administration of NS-065/NCNP-01: A phase 1, dose escalation, first-in-human study**
H. Komaki; T. Nagata; T. Saito; S. Masuda; E. Takeshita; H. Tachimori; M. Sasaki; S. Takeda

G.P.252 **Idebenone improves respiratory function in pediatric and adolescent patients with Duchenne muscular dystrophy not using glucocorticoid steroids**
G. Buyse

G.P.253 **CAT-1004. an oral agent targeting NF-kB in development for treatment of Duchenne muscular dystrophy: Phase 1/2 study design**
J. Donovan; H. Sweeney; K. Vandenborne; B. Russman; M. Jirousek; R. Finkel

G.P.254 **VISION – DMD: drug development of VBP15, an experimental steroid-like drug for DMD**
P. Clemens; M. Guglieri; A. Cnaan; J. Damsker; H. Gordish-Dressman; L. Morgenroth; K. Nagaraju; Y. Hathout; E. Hoffman; K. Bushby; L. Morgenroth Hache

G.P.255 **HT-100: Interim safety, pharmacokinetics, and pharmacodynamic data in DMD boys**
D. Escolar; M. Blaustein; E. Bush; K. Wagner; B. Wong; K. Flanigan; J. Han; A. Britton; J. Loewy; K. Dykstra; A. Connolly

G.P.256 **Withdrawn**

G.P.257 **Nonclinical pharmacokinetic evaluation of Eteplirsen, SRP-4045, and SRP-4053; Three Phosphorodiamidate morpholino oligomers (PMOs) for the treatment of patients with Duchenne muscular dystrophy (DMD)**
P. Sazani; J. Charleston; C. Shanks; J. Zhang; M. Carver; J. Saoud; E. Kaye

G.P.258 **Eteplirsen, a Phosphorodiamidate morpholino oligomer (PMO) for the treatment of Duchenne muscular dystrophy (DMD): Clinical update**
E. Kaye; J. Mendell; L. Rodino-Klapac; Z. Sahenk; L. Lowes; L. Alfano; K. Berry; A. Gomez Ramirez; S. Lewis; K. Flanigan; L. Cripe; S. Al-Zaidy; P. Duda; P. Sazani; J. Saoud

G.P.259 **A novel clinical trial design to evaluate the efficacy and safety of two exon-skipping PMOs, SRP-4045 and SRP-4053, in patients with Duchenne muscular dystrophy**
G. Laforet; E. Kaye; J. Saoud

G.P.260 **Are effects of Translarna (ataluren) on muscle strength more discernable in younger patients with Duchenne muscular dystrophy (DMD)?**
C. McDonald; A. Reha; G. Elfring; P. Trifilis; S. Park; T. Ong; S. Peltz; R. Spiegel; Ataluren DMD Study Group

Congenital muscular dystrophies (G.P.261–282)
Facilitators: Carsten Bönneman and Anna Sarkozy

G.P.261 **The application of antisense oligonucleotide therapy in Collagen 6-related congenital muscular dystrophy**
H. Zhou; E. Marrosu; F. Muntoni

G.P.262 **Early scoliosis surgery may prevent deterioration of respiratory function in Ullrich congenital muscular dystrophy**
K. Kohashi; A. Ishiyama; E. Takeshita; Y. Shimizu-Motohashi; T. Saito; E. Nakagawa; H. Komaki; K. Sugai; I. Nishino; W. Saito; M. Takaso; M. Sasaki

G.P.263 **Comparison of upper extremity measures in individuals with COL6 and LAMA2-muscular dystrophies**
M. Jain; K. Meilleur; C. Nichols; M. Waite; R. Parks; B. Hodsdon; R. Bendixen; N. Hsia; A. Glanzman; L. Nelson; K. Keller; T. Duong; M. McGuire; J. Dastgir; S. Donkervoort; M. Leach; J. Collins; C. Vuillerot; A. Rutkowski; C. Bönneman

G.P.264 **Electrical impedance myography as a potential biomarker in individuals with COL6-related dystrophy**
C. Nichols; T. Lehky; M. Waite; T. Duong; L. Nelson; K. Keller; D. Lott; K. Meilleur; J. Collins; J. Dastgir; C. Vuillerot; A. Rutkowski; S. Donkervoort; M. Leach; M. Jain; C. Bönneman

G.P.265 **Validation of actiGraph GT3X accelerometers in collagen 6-related muscular dystrophy and LAMA2-related muscular dystrophy**
K. Meilleur; J. Elliott; M. Linton; C. Vuillerot; R. Bendixen; I. Arveson; F. Tounkara; M. Waite; C. Nichols; K. Yang; S. Donkervoort; J. Dastgir; M. Leach; C. Bönneman; M. Jain

G.P.266 **Gait in children with collagen 6 disorders – A prospective study**
K. Carroll; R. Kennedy; K. de Valle; M. Ryan; A. Kornberg

G.P.267 **Clinical, pathologic, genetic features of collagen 6-related myopathy in Korea**
J. Lee; H. Park; Y. Park; S. Kim; Y. Choi

G.P.268 **Col6A2 null mice are a new mouse model of collagen-VI related dystrophies and relevant to the human disease**
P. Mohassel; J. Rooney; Y. Zou; C. Bönnemann

G.P.269 **Correlation between crural muscle fatty change and maximum motor function in Fukuyama congenital muscular dystrophy patients**
K. Ishigruo; K. Ishigaki; T. Sato; T. Murakami; M. Shichiji; K. Saito; M. Osawa; S. Nagata

G.P.270 **Assessment of growth and development in patients with Fukuyama congenital muscular dystrophy using bioelectrical impedance analysis**
T. Murakami; T. Uchiyama; T. Nakayama; K. Ishiguro; T. Sato; S. Kuru; K. Ishigaki

G.P.271 **The gross motor function measure is a valid scale for assessing Fukuyama congenital muscular dystrophy**
T. Sato; K. Ishigaki; M. Zushi; M. Adachi; K. Goto; M. Akiduki; K. Ishiguro; M. Shichiji; T. Murakami; K. Saito; M. Osawa; T. Ikai; I. Kondo; S. Nagata

G.P.272 **Eight novel UK families further expand current knowledge on GMPPB-gene related dystroglycanopathies**
A. Sarkozy; S. Torelli; R. Barresi; M. Bertoli; M. Sframeli; R. Mein; M. Yau; C. Sewry; R. Phadke; L. Feng; P. Ala; A. Manzur; K. Bushby; H. Lochmüller; T. Willis; F. Norwood; R. Rakowicz; F. Muntoni

G.P.273 **Homozygous splicing mutation in ISPD gene in a girl with Walker–Warburg syndrome**
J. Tanboon; S. Noguchi; S. Mitsuhashi; I. Nishino

G.P.274 **DAG1 mutations associated with asymptomatic hyperCKemia and hypoglycosylation of α-dystroglycan**
S. Noguchi; M. Dong; Y. Endo; Y. Hayashi; S. Yoshida; I. Nonaka; I. Nishino

G.P.275 **POMGNT2 mutations are associated with milder forms of limb girdle muscular dystrophy**
Y. Endo; S. Noguchi; M. Dong; M. Ogawa; Y. Hayashi; S. Kuru; K. Sugiyama; S. Nagai; S. Ozasa; I. Nonaka; I. Nishino

G.P.276 **Clinical summary of eight unrelated individuals with GMPPB mutations**
K. Mathews; B. Jensen; D. Saade; T. Willer; M. Cox; T. Mozaffar; M. Scavina; T. Winder; K. Campbell; S. Moore

G.P.277 **Diagnostic difficulties in muscular dystrophy due to LAMA2 mutations – A case report of affected father and his son**
J. Haberlová; R. Barresi; L. Fajkusová

G.P.278 **Next generation sequencing (NGS): A powerful tool for studying rigid spine patients and multiminicore myopathy**
J. Gurgel-Giannetti; E. Concentino; M. Lazar; V. Van der Linden; A. Giannetti; F. Fernandes; W. Campos; M. Vainzof

G.P.279 **Congenital muscular dystrophies in the UK population: Update of clinical and molecular spectrum of patients diagnosed over a 12-year period**
M. Sframeli; A. Sarkozy; M. Bertoli; G. Astrea; J. Hudson; M. Scoto; R. Mein; M. Yau; R. Phadke; L. Feng; C. Sewry; S. Robb; A. Manzur; S. Messina; K. Bushby; F. Muntoni

G.P.280 **Relative prevalence of congenital muscular dystrophy subtypes in Korea**
A. Cho; B. Lim; K. Kim; Y. Hwang; J. Chae

G.P.281 **Combined cell and gene therapy to treat merosin deficient congenital muscular dystrophy**
D. Velardo; T. Domi; E. Porrello; A. Capotondo; A. Biffi; R. Tonlorenzi; S. Takeda; S. Amadio; M. Ruegg; S. Previtali

G.P.282 **Launching the first clinical trial in SEPN1-related myopathy: The SELNAC study**
C. Dill; H. Prigent; A. Behin; F. Piemonte; E. Bertini; D. Orlikowski; B. Estournet; A. Ferreiro

General neuromuscular (G.P.283–293)
Facilitators: Jan Kirschner and Max Damian

G.P.283 **Utility of a respiratory database in a tertiary neuromuscular clinic; a 10-year experience**
T. Mozaffar; N. Goyal; T. Cash; S. Rai; A. Wang; L. Chui

G.P.284 **The TREAT-NMD Advisory Committee for Therapeutics (TACT): An innovative de-risking model to foster orphan drug development**
D. Wells; E. Heslop; C. Csimma; V. Straub; J. McCall; K. Nagaraju; K. Wagner; D. Caizergues; R. Korinthenberg; K. Flanigan; J. Mendell; M. Kelly; P. Kaufmann; E. McNeil; A. Robertson; L. Johnston; K. Bushby

G.P.285 **TREAT-NMD (translational research in Europe, assessment and treatment for neuromuscular disorders)**
A. Aartsma-Rus; E. Hoffman; F. Bucella; K. Flanigan; J. Kirschner; A. Kole; E. Mercuri; I. Nishino; K. North; A. Pereda Alonso; J. Rahbek; T. Sejersen; M. Wood; S. Lynn; A. Robertson; H. Lochmüller; V. Straub; K. Bushby

G.P.286 **Developing digital tools for improving patient and family engagement in paediatric clinical research**
O. Veldhuizen; C. Turner; M. Hails; V. Straub

G.P.287 **ScanBank – The development of an online database for neuromuscular MRI scans**
O. Veldhuizen; C. Wood; A. Murphy; R. Thompson; N. Steen Krogh; V. Straub

G.P.288 **Characteristic MRI findings of upper limb muscle involvement in myotonic dystrophy type 1**
K. Sugie; M. Sugie; T. Taoka; Y. Tonomura; A. Kumazawa; T. Izumi; S. Ueno; K. Kichikawa

G.P.289 **Validation of a two-component EPG-model to estimate the muscle T2 water values by 1H-MRS**
H. Reyngoudt; P. Baudin; B. Marty; N. Azzabou; E. Caldas de A. Araujo; P. Loureiro de Sousa; P. Carlier

G.P.290 **Paraspinal fat infiltration in healthy adults with aging**
J. Dahlqvist; C. Vissing; G. Hedermann; C. Thomsen; J. Vissing

G.P.291 **Assessment of the 2- and 6-minute walk test for patients with neuromuscular diseases**
L. Andersen; K. Knak; N. Witting; J. Vissing

G.P.292 **Test-retest reliability of the 2- and 6-minute walk tests in patients with neuromuscular diseases**
K. Knak; L. Andersen; N. Witting; J. Vissing

G.P.293 **Evaluating the benefits of community based aerobic training on the physical health and well-being of people with neuromuscular diseases: A pilot study**
A. Wallace; L. Dewar; A. Pietrusz; M. Dudziec; A. Sterr; M. Laura; I. Skorupinska; M. Skorupinska; M. Hanna; M. Trenell; G. Baio; M. Reilly; G. Ramdharry

Congenital myopathies 1 (G.P.294–308)
Facilitators: Norma Romero and Heinz Jungbluth

G.P.294 **Peripheral vein injection of AAV8-MTM1 leads to long-term survival and correction of severe muscle pathology in a canine model of X-linked myotubular myopathy: Results from a dose escalation study**
D. Mack; K. Poulard; M. Goddard; J. Snyder; R. Grange; J. Doering; J. Strande; V. Latournerie; P. Veron; L. Yang; L. Buscara; C. Le Bec; S. Martin; M. O'Callaghan; F. Mingozzi; A. Beggs; M. Lawlor; F. Mavilio; M. Childers; A. Buj-Bello

G.P.295 **X-linked myotubular myopathy in females**
C. Fiorillo; F. Fattori; G. Astrea; M. Pedemonte; A. Rubegni; F. Trucco; A. Tessa; M. Savarese; J. Baldacci; P. Broda; E. Bertini; C. Minetti; V. Nigro; C. Bruno; F. Santorelli

G.P.296 **Natural history and functional status of patients with myotubular myopathy enrolled in a prospective and longitudinal study**
N. R'guiba; M. Annoussamy; R. Cardas; C. Lilien; G. Ollivier; F. Muntoni; C. Bönnemann; V. Biancalana; J. Cuisset; M. Mayer; H. Landy; D. Ramsdell; M. Nelken; A. Le Moing; T. Gidaro; F. Mingozzi; A. Buj-Bello; J. Hogrel; T. Voit; L. Servais

G.P.297 **Dominant BIN1-related centronuclear myopathy (CNM) revealed by lower limb myalgia and moderate CK elevation**
M. Garibaldi; N. Romero; J. Böhm; P. Ottaviani; F. Fattori; F. Laschena; J. Laporte; E. Bertini; G. Antonini

G.P.298 **Non-invasive NMR study of the mouse model for centronuclear myopathy with mutation in the dynamin-2 gene**
A. Martins Bach; B. Matot; C. Wary; M. Bitoun; M. Vainzof; P. Carlier

G.P.299 **A family with DNM2-related centronuclear myopathy without ophthalmoplegia**
J. Park; S. Kim; D. Kim; J. Shin

G.P.300 **Recessive loss-of-function SCN4A mutations associated with a novel phenotype of congenital myopathy**
I. Zaharieva; M. Thor; E. Oates; C. Karnebeek; E. Kamsteeg; L. Hartley; E. Blom; N. Witting; M. Rasmussen; M. Gabbett; G. Ravenscroft; M. Hanna; P. Ruben; S. Lewis; R. Mannikko; F. Muntoni; SCN4A Research Group

G.P.301 **Novel myopathy in a new-born with severe thoracic dysplasia caused by mutations in the SBDS gene. Further delineation of the phenotypic spectrum of Shwachman–Diamond syndrome**
C. Hedberg Oldfors; A. Topa; M. Tulinius; A. Oldfors

G.P.302 **Late-onset zebra body myopathy**
D. Pehl; J. Radke; K. Irlbacher; F. Heppner; H. Goebel; W. Stenzel

G.P.303 **A novel mutation in PIEZO2 in a family presenting with autosomal dominant myopathy, ptosis. external ophthalmoplegia and distal symphalangism**
M. Bertoli; A. Topf; E. Harris; S. Laval; A. Sarkozy; H. Lochmüller; S. Lynch; V. Straub

G.P.304 **WES revealed a de-novo missense mutation in the NALCN gene in a Freeman–Sheldon-(DA2A) like syndrome with CNS involvement**
M. Karakaya; V. Kunde; R. Heller; P. Nürnberg; S. Cirak

G.P.305 **Surgical correction of scoliosis in patients with congenital myopathies**
F. Trucco; A. Andaloro; M. Pedemonte; V. Lanzillotta; C. Panicucci; C. Fiorillo; F. Becchetti; C. Minetti; C. Bruno

G.P.306 **Late-onset congenital myopathies: Clinical and molecular features**
L. Maggi; I. Colombo; F. Fattori; C. Fiorillo; G. Vita; F. Magri; M. Pane; A. D'Amico; P. Bernasconi; M. Mora; S. Messina; F. Santorelli; E. Mercuri; E. Bertini; E. Pegoraro; G. Comi; M. Moggio; L. Morandi; C. Bruno

G.P.307 **Congenital myopathies: Characteristics and subtypes in Hong Kong**
S. Chan; R. Ho; A. Chan; J. Ip; S. Wong; G. Ng; H. Lee; Y. Cheng; K. Liu; C. Lee; T. Fung; W. Cherk; S. Chan; W. Lam; W. Shek; V. Wong

G.P.308 **Congenital myopathy, obesity, overgrowth, distinctive features and lymphoedema in a family; chance association or overgrowth syndrome**
P. Dawson; B. Toms; C. Wragg; A. Majumdar; K. Vijayakumar; S. Love

15:45 **Afternoon refreshments –** Corn Exchange, café and mezzanine

16:30–18:00 **Guided poster discussion session 4: Parallel sessions (G.P.309–418) –** Corn Exchange

EDMD and distal myopathies (G.P.309–323)
Facilitators: Peter Hackmann and Carina Wallgren-Pettersson

G.P.309 **Muscle MRI findings in myopathies due to FHL1 mutation**
P. Mohassel; S. Donkervoort; J. Dastgir; F. Munell; A. Sanchez; J. Haverlova; A. Manzur; S. Quijano-Roy; G. Tasca; B. Udd; F. Muntoni; C. Bönnemann

G.P.310 **Late-onset mild myopathy with protein aggregates in two transgenic mouse models of FHL1**
J. Rooney; Y. Zou; B. Cowling; C. Mitchell; C. Bönnemann

G.P.311 **A novel LMNA mutation causes severe congenital phenotype with cytoplasmic bodies**
A. Nishikawa; S. Mitsuhashi; H. Mitsuhashi; A. Uruha; S. Noguchi; I. Nishino

G.P.312 **Overlap laminopathy with mild neurogenic atrophy and overt muscular dystrophy**
N. Voermans; B. Kusters; F. Megen; J. Jongbloed; N. Alfen; J. Verschuuren; B. van Engelen; B. Udd

G.P.313 **Steroid benefit in a laminopathy-congenital muscular dystrophy patient with dropped head syndrome: A 10-year follow-up**
J. Gurgel-Giannetti; B. Ribeiro; L. Uliana; G. Sampaio; A. Giannetti; V. Van der Linden; M. Vainzof

G.P.314 **Gene therapy via trans-splicing for LMNA-related congenital muscular dystrophy (L-CMD)**
F. Azibani; L. Arandel; M. Beuvin; A. Jollet; B. Prudhon; S. Lorain; A. Bertrand; G. Bonne

G.P.315 **Coexisting TIA1 and MYH7 distal myopathy**
P. Brand; P. Dyck; J. Liu; S. Berini; D. Selcen; M. Milone

G.P.316 **The FINmaj mutation in TTN induces the loss of several protein domains from titin C-terminus**
A. Vihola; K. Charton; J. Sarparanta; P. Jonson; I. Richard; B. Udd

G.P.317 **Oculopharyngodistal myopathy in a Korean family**
J. Shin; D. Kim; K. Lee; J. Sung

G.P.318 **Longitudinal study for GNE gene (ClinBio-GNE)**
T. Gidaro; D. Duchene; G. Ollivier; V. Decostre; J. Hogrel; P. Carlier; A. Behin; L. Servais

G.P.319 **GNE myopathy world-wide epidemiology based on the patient self-reported registry**
O. Pogoryelova; P. Cammish; A. Skrinar; S. Rao; H. Lochmüller; E. Kakis

G.P.320 **MYH7-related myopathies: Clinical. histopathological and imaging findings in a cohort of Italian patients**
C. Fiorillo; M. Savarese; G. Astrea; D. Cassandrini; L. Ruggiero; M. Fanin; L. Vercelli; A. D'Amico; M. Pane; G. Tasca; M. Morandi; E. Pegoraro; L. Santoro; E. Mercuri; M. Mora; E. Bertini; C. Minetti; F. Santorelli; V. Nigro; C. Bruno

G.P.321 **Analysis of the pathogenesis of vocal cord and pharyngeal weakness with distal myopathy**
S. Yamashita; T. Nishikami; N. Tawara; T. Doki; Y. Nakajo; Y. Matsuo; Y. Yonemochi; M. Nagai; Y. Maeda; Y. Ando

G.P.322 **Unique myopathy presenting in adulthood with proximal muscle weakness and respiratory insufficiency**
N. Goyal; M. Waldrop; T. Mozaffar

G.P.323 **Targeted next-generation sequencing reveals novel TTN mutations causing recessive distal titinopathy**
A. Evilä; S. Penttilä; P. Hackman; B. Udd

HMSN (G.P.324–333)
Facilitators: Mary Reilly and Gita Ramdharry

G.P.324 **Neuropathological findings from a human post mortem case of distal hereditary motor neuropathy (dHMN) due to p.Ser135Phe HSPB1 mutation and transgenic mice with mutant or wild-type HSP27 overexpression**
R. Phadke; A. Rossor; V. Benoy; B. Kalmar; R. King; L. Greensmith; L. Bosch; M. Reilly; H. Houlden

G.P.325 **Gait variability during prolonged walking in children and adolescents with Charcot–Marie–Tooth disease**
R. Kennedy; K. Carroll; M. Ryan; J. McGinley

G.P.326 **Deletion of the P2 promoter of the GJB1 gene confirms X-linked Charcot–Marie–Tooth disease in a large family**
R. Kulshrestha; T. Antoniadi; S. Burton-Jones; M. Rogers; N. Kiely; T. Willis

G.P.327 **Dynamic pedobarography assessment in children and adolescents with Charcot–Marie–Tooth disease**
A. Mattiello-Sverzut; A. Nascimento-Elias; C. Baptista; P. Calori; B. Garcia; C. Sartor; I. Sacco; W. Marques Jr

G.P.328 **Stabilometric findings in children and adolescent with Charcot–Marie–Tooth Disease**
A. Mattiello-Sverzut; C. Baptista; P. Calori; B. Garcia; W. Marques Jr

G.P.329 **Giant axonal neuropathy – Clinical trial preparedness and evaluation of markers of disease severity**
D. Bharucha-Goebel; D. Ezzo; M. Jain; M. Waite; C. Nichols; T. Lehky; P. Mohassel; S. Donkervoort; M. Leach; J. Dastgir; J. Marra; W. Zein; C. Bönnemann

G.P.330 **Comparison of two clinical motor scales in individuals with giant axonal neuropathy (GAN)**
M. Waite; D. Bharucha-Goebel; T. Moulton; C. Zampieri; C. Nichols; K. Alter; G. Averion; S. Donkervoort; M. Leach; J. Dastgir; P. Mohassel; M. Jain; C. Bönneman

G.P.331 **A novel 9 amino acid in-frame deletion in the NTRK1 tyrosine kinase domain in a patient with congenital insensitivity to pain with anhidrosis (CIPA)**
N. Forrester; S. Burton-Jones; T. Antoniadi; A. Norman; A. Majumdar; K. Vijayakumar

G.P.332 **Charcot–Marie–Tooth type 4B1 (MTMR2 gene): Confounding clinical presentation and report of 5 original mutations**
R. Guimarães-Costa; P. Latour; X. Ferrer; G. Solé; I. Husson; A. Lacour; O. Dubourg; S. Leonard-Louis; T. Stojkovic

G.P.333 **Normative aerobic exercise values in CMT**
A. Wallace; L. Dewar; A. Sterr; M. Hanna; M. Trenell; A. Pietrusz; M. Dudziec; P. Hennis; R. Stokes; M. Reilly; G. Ramdharry

Congenital myopathies 2 (G.P.334–346)
Facilitators: Kristen Nowak and Kathryn North

G.P.334 **Etiological yield of muscle biopsy in the newborn period**
E. Serdaroglu; G. Haliloglu; B. Talim; S. Yigit; M. Yurdakok; H. Topaloğlu

G.P.335 **Molecular basis of stiff patient syndrome caused by mutations in ACTA1 and TPM3**
M. Papadaki; S. Marston; M. Memo; A. Messer; S. Donkervoort; C. Bönnemann; K. Nowak; R. Ong; E. McNamara

G.P.336 **Cytoplasmic body pathology in severe ACTA1-myopathy in the absence of typical nemaline-rod histology**
S. Donkervoort; S. Chan; N. Bradley; A. Foley; D. Nguyen; Y. Hu; M. Leach; M. Thangarajh; C. Reyes; J. Nance; S. Moore; C. Bönnemann

G.P.337 **Cardiomyopathy in patients with ACTA1-myopathy**
S. Donkervoort; M. Yang; M. Leach; L. Medne; S. Yum; L. Hotchkiss; A. Rutkowski; K. Chatfield; S. Auerbach; S. Miyamoto; C. Bönnemann

G.P.338 **A novel form of cap myopathy in absence of heart disease associated with recessive TTN gene mutations**
C. Jou; C. Ortez; C. Jimenez-Mallebrera; N. Romero; G. Bouchier; A. Fernández-Marmiesse; M. Couce; M. Carrascosa-Romero; A. Blancas; T. Ribalta; J. Colomer; A. Nascimento

G.P.339 **Highly variable skeletal muscle histo-immunocytochemical and ultrastructural features in titin-related myopathies**
R. Avila-Polo; E. Malfatti; I. Nelson; J. Nectoux; J. Böhm; O. Abath-Neto; B. Eymard; S. Monges; F. Lubieniecki; G. Brochier; M. Beuvin; A. Madelaine; C. Labasse; A. Taratuto; B. Udd; I. Richard; F. Leturcq; G. Bonne; J. Laporte; N. Romero

G.P.340 **Nemaline myopathy: Next generation sequencing (NGS) significantly improving the molecular classification of Brazilian families**
J. Gurgel-Giannetti; M. Lazar; R. Pavanello; E. Concentino; F. Fernandes; G. Sampaio; M. Zatz; M. Vainzof

G.P.341 **Functional assessment of nebulin missense variants**
J. Laitila; M. Marttila; V. Lehtokari; C. Wallgren-Pettersson; K. Pelin

G.P.342 **Novel histopathological phenotypes associated with TPM2 and TPM3-related congenital myopathies**
E. Malfatti; O. Abath Neto; M. Fardeau; J. Laporte; N. Romero

G.P.343 **Genetic profiles in patients with nemaline myopathy**
J. Lee; H. Jang; S. Kim; J. Shin

G.P.344 **Mutations in the EF hands of STIM1 lead to different clinical severity**
E. Dionnet; P. Tsvetkov; S. Gorokhova; A. Maues de Paula; F. Devred; M. Krahn; M. Bartoli

G.P.345 **A novel STIM1 mutation at p.340 causes tubular aggregate myopathy with miosis without additional features of Stormorken syndrome**
E. Harris; J. Hudson; J. Marsh; C. Marini Bettolo; M. Neri; A. Ferlini; K. Bushby; H. Lochmüller; V. Straub; R. Barresi

G.P.346 **Muscle imaging in STIM1-mutated tubular aggregate myopathy patients**
G. Tasca; A. D'Amico; M. Monforte; A. Nadaj-Pakleza; M. Vialle; F. Fattori; J. Vissing; E. Ricci; E. Bertini

***mdx* mouse** (G.P.347–366)
Facilitators: Sue Brown and George Dickson

G.P.347 **Long term follow-up of GRMD dogs transplanted with human adipose derived stem cells**
M. Zatz; J. Gomes; M. Pelatti; M. Secco; N. Vieira; E. Zucconi; M. Vainzof; V. Landini; T. Andrade

G.P.348 **Gene delivery to large animal models using AAV vectors**
J. Chamberlain; J. Seto; J. Ramos; S. Hauschka; G. Odom

G.P.349 **Lack of estrogens aggravates the pathology in dystrophic mice**
E. Gayi; H. Ismail; L. Neff; O. Patthey-Vuadens; T. Saibara; K. Toda; U. Ruegg; L. Scapozza; O. Dorchies

G.P.350 **Sarcolemmal nNOSμ targeting in *mdx* mice improves muscle function and restores dystrophin complex proteins via α-syntrophin**
M. Kim; D. Rebolledo; N. Whitehead; M. Adams; S. Froehner

G.P.351 **Effect of different dosing regimens of exon skipping antisense oligonucleotides on functional outcome in the *mdx* mouse model of Duchenne muscular dystrophy**
N. Datson; R. Weij; S. Bijl; A. Janson; R. Vermue; J. Testerink; J. van Deutekom

G.P.352 **The *mdx* mutation in the 129/Sv background results in a milder phenotype: Transcriptome comparative analysis searching for the protective factors**
P. Calyjur; C. Almeida; A. Santos; P. Onofre-Oliveira; M. Vainzof

G.P.353 **Effect of treatment with grow factors on muscle pathology in the *mdx* mouse model of Duchenne muscular dystrophy**
T. Nielsen; T. Pinos; T. Krag; J. Vissing

G.P.354 **NHE1 inhibition as a potential therapeutic strategy to attenuate DMD pathology**
P. Ioannou; U. Burki; S. Laval; S. Schaefer; V. Straub

G.P.355 **Adiponectin, a powerful brake on skeletal muscle inflammation and injury**
M. Abou-Samra; S. Lecompte; O. Schakman; L. Noel; A. Lafosse; M. Many; P. Gailly; S. Brichard

G.P.356 **Preclinical data for a novel Toll-like receptor antagonist in *mdx* mice support its clinical development as a potential treatment for disease-related muscle inflammation in Duchenne muscular dystrophy**
J. Boehler; K. Nagaraju; M. Hurtt; S. Agrawal

G.P.357 **Early expression of ΔCH1 dystrophin isoform reverses or prevents muscular dystrophy in the Dup2 mouse**
N. Wein; A. Vulin; T. Simmons; A. Molza; F. Gumienny; N. Huang; O. Delalande; J. Ervasti; R. Weiss; K. Flanigan

G.P.358 **The *mdx*^{5Cv} dystrophic mouse: In depth longitudinal phenotyping**
O. Dorchies; E. Gayi; H. Ismail; L. Neff; O. Patthey-Vuadens; L. Scapozza; U. Ruegg

G.P.359 **Ablation of MMP-9 promotes resolution of inflammation and regeneration by modulating chemotaxis in early stage but exacerbates fibrosis in late stage in *mdx* mice**
N. Shiba; D. Miyazakaki; T. Yoshizawa; K. Fukushima; M. Imamura; S. Takeda; A. Nakamura

G.P.360 **Neuromuscular synapse characteristics of mice with low dystrophin levels**
E. van der Pijl; M. van Putten; J. Verschuuren; A. Aartsma-Rus; J. Plomp

G.P.361 **Phenotypic differences in *mdx* mice on C57Bl/10 and DBA/2 backgrounds**
S. Keenan; B. Snyder; H. Xiao; A. Robertson; M. Ahlijanian; L. Bristow; N. Devidze

G.P.362 **Effects of ready-to-use compounds with different actions on mechano-sensitive oxidative stress in exercised *mdx* mice**
P. Mantuano; A. Cozzoli; R. Capogrosso; A. Massari; L. De Benedictis; M. Montagnani; G. Camerino; A. Giustino; A. De Luca

G.P.363 **Glucose uptake and mitochondrial function following 8 weeks of dietary nitrate supplementation in the dystrophin-deficient *mdx* mouse**
C. Timpani; A. Trewin; A. Betik; N. Stepto; A. Hayes; G. McConell; E. Rybalka

G.P.364 **D-amino acid substitution of peptide-mediated NF-κB suppression in *mdx* mice preserves therapeutic benefit in skeletal muscle. but causes kidney toxicity**
P. Clemens; D. Reay; S. Bastacky; K. Wack; D. Stolz; P. Robbins

G.P.365 **Effect of forced and voluntary exercise on muscle function and integrity in aged mice expressing low dystrophin levels**
M. van Putten; B. Kogelman; R. Werring; C. Tanganyika-de Winter; L. van der Weerd; A. Aartsma-Rus

G.P.366 **Effect of forced and voluntary exercise on heart function in mice with low dystrophin levels**
M. van Putten; B. Kogelman; C. Tanganyika-de Winter; R. Werring; M. Hulsker; L. van der Weerd; A. Aartsma-Rus

Next generation sequencing (G.P.367–384)
Facilitators: Giorgio Tasca and Nigel Laing

G.P.367 **Muscle MRI correlates with histology and clinical features in patients with FSHD, OPMD and sIBM**
S. Lassche; M. Schyns; N. Koster; L. Heskamp; A. Heerschap; B. Kusters; N. Voermans; B. van Engelen

G.P.368 **Accelerated multi spin echo NMR imaging for muscle water T2 and fat fraction determination with compressed sensing**
B. Coppa; B. Marty; P. Baudin; N. Azzabou; P. Carlier

G.P.369 **Quantitative MRI and MRS detect alterations in muscle quality in both congenital muscular dystrophy and Duchenne muscular dystrophy**
R. Willcocks; W. Triplett; D. Lott; S. Forbes; J. Dastgir; C. Bönneman; K. Vandenborne; G. Walter

G.P.370 **Simultaneous muscle water T2 and fat fraction mapping from NMR transverse relaxometry with stimulated echo compensation**
B. Marty; P. Baudin; H. Reyngoudt; N. Azzabou; E. Araujo; P. Carlier; P. de Sousa

G.P.371 **Skeletal muscle tissue characterization by ^{23}Na NMRS under different vascular filling conditions**
B. Marty; T. Gerhalter; E. Araujo; E. Giacomini; P. Carlier

G.P.372 **Caveolinopathy presenting with muscle pain and rhabdomyolysis**
R. Scalco; A. Gardiner; R. Pitceathly; C. Turner; M. Parton; D. Fialho; M. Hanna; H. Houlden; J. Holton; A. Manzur; F. Lucy; D. Hilton-Jones; A. Schapira; E. Murphy; R. Barresi; H. Jungbluth; R. Phadke; R. Quinlivan

G.P.373 **Large screening of patients diagnosed as limb girdle muscular dystrophy or congenital myopathy using Motorplex**
M. Savarese; G. Di Fruscio; C. Bruno; A. Torella; T. Giugliano; M. Mora; L. Morandi; G. Siciliano; A. Toscano; A. Garofalo; T. Mongini; C. Angelini; L. Santoro; K. Claes; E. Bertini; F. Santorelli; G. Comi; S. Sacconi; L. Politano; V. Nigro

G.P.374 **High-throughput genetic testing for muscle disease and an exome database of the undiagnosed in Japan**
S. Mitsuhashi; A. Nishikawa; W. Zhu; T. Termglinchan; J. Shin; K. Takayama; A. Uruha; Y. Endo; S. Noguchi; I. Nishino

G.P.375 **Novel variant blossom: From pathology to next generation sequencing to cellular biology**
W. Zhu; S. Mitsuhashi; W. Liang; T. Ito; I. Nishino

G.P.376 **Collaboration in NeurOmics: Enabling effective data-sharing and maximising impact in neuromuscular disease**
C. Turner; K. Bushby; L. Johnston; H. Lochmüller; O. Riess; B. Wirth; V. Straub; R. Thompson; G. van Ommen

G.P.377 **NeurOmics: EU-funded-omics research for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases**
C. Turner; A. Brice; K. Bushby; O. Riess; M. Hanna; G. van Ommen; F. Muntoni; T. Klockgether; B. Wirth; H. Lochmüller; V. Timmerman; L. Schoells; V. Straub; S. Tabrizi

G.P.378 **Omics approach and novel biostatistic tools identified RPL3L as potential genetic modifier of clinical severity in female carriers of Duchenne muscle dystrophy**
M. Neri; C. Scotton; C. Scapoli; A. Carrieri; F. Di Raimo; M. Bovolenta; S. Gherardi; A. Armaroli; C. Passarelli; A. D'Amico; E. Bertini; M. Pane; E. Mercuri; G. Pesole; L. Wenyan; F. Mingyan; F. Gualandi; E. Schwartz; A. Yuryev; A. Ferlini

G.P.379 **Whole exome sequencing at the Institute of myology in the context of the myocapture project to identify novel genes of myopathies**
I. Nelson; R. BenYaou; J. Nectoux; C. Masson; F. Leturq; P. Richard; T. Stojkovic; A. Behin; P. Laforêt; V. Allamand; B. Eymard; G. Bonne

G.P.380 **Efficacy of next-generation sequencing in molecular diagnosis of archived DNA samples**
S. Beecroft; R. Ong; K. Yau; R. Duff; R. Allcock; M. Davis; P. Lamont; N. Laing

G.P.381 **Targeted next-generation sequencing approach for molecular diagnosis of inherited muscular disorders**
H. Park; J. Lee; Y. Choi

G.P.382 **Filling in the gap between exome and genome: mRNA analysis as a clinical diagnosis tool**
H. Gonorazky; G. Yoon; N. Sabha; E. Tsuchiya; K. Amburgey; C. Marshall; J. Dowling

G.P.383 **Dystrophinopathies: A NGS approach for the molecular analysis of DMD gene**
P. Gallano; L. Gonzalez-Quereda; A. Lasa; M. Baena; M. Rodriguez; S. Monges; F. Lubieniecki; A. Nascimento; C. Ortez; M. Baiget

G.P.384 **Use of next generation exome sequencing for the diagnosis of patients with pediatric neuromuscular disorders**
F. Munell; S. Ferrer; A. Sánchez-Montáñez; E. Martínez-Sáez; M. Gractacós; M. Pérez; E. Cuenca; M. Roig-Quilis; M. Olivé; F. Vidal

DMD clinical (G.P.385–404)
Facilitators: Brenda Wong and Giles Campion

G.P.385 **Strategies for engaging the Duchenne muscular dystrophy community in research**
P. Clemens; L. Morgenroth; K. Clinard; R. Bendixen

G.P.386 **DMD registry in Turkey: Highlights**
H. Topaloğlu; G. Haliloglu; O. Yilmaz; A. Karaduman

G.P.387 **Overview of morbidity. mortality and survival in Duchenne muscular dystrophy in the North East of England**
C. Marini Bettolo; H. van Ruiten; M. Guglieri; M. Eagle; V. Straub; H. Lochmüller; K. Bushby

G.P.388 **Health related quality of life in European adults with DMD: Results from the Care-NMD-project**
B. Steffensen; C. Otto; U. Werlauff; J. Rahbek; A. Hoejberg; J. Kirschner; J. Vry; K. Gramsch

G.P.389 **A prospective natural history study to measure progression of physical impairment. activity limitation and quality of life in Duchenne muscular dystrophy**
N. Goemans; B. Wong; C. McDonald; C. Mason; A. Hall; G. Campion

G.P.390 **Vitamin D in corticosteroid treated Duchenne muscular dystrophy: What dose achieves serum 25OH vitamin D sufficiency?**
N. Alshaikh; A. Brunklaus; T. Davis; S. Robb; R. Quinlivan; P. Munot; A. Sarkozy; F. Muntoni; A. Manzur

G.P.391 **Clinical outcomes in a large cohort of boys and adolescents with Duchenne muscular dystrophy**
N. Alshaikh; A. Brunklaus; S. Robb; R. Quinlivan; P. Munot; A. Sarkozy; F. Muntoni; A. Manzur

G.P.392 **Five-year longitudinal UC Davis CINRG Duchenne Natural History (DNHS) Study data shows mobility-focused POSNA PODCI items are sensitive to 12-month disease progression across all stages of DMD functional ability**
E. Henricson; C. McDonald; The CINRG Investigators

G.P.393 **Kinesthesia in children with Duchenne muscular dystrophy**
B. Akel; S. Karayazgan; S. Subası; N. Bulut; O. Yılmaz; A. Karaduman

G.P.394 **Physical activity in boys with DMD is lower and less demanding compared to healthy boys**
L. Heutinck; N. van Kampen; M. Jansen; I. de Groot

G.P.395 **Does an inability to achieve plantar grade at the ankle joint have an impact on the outcome measures of the six minute walking test and North Star ambulatory assessment; and is this further compounded by ilio-tibial band tightness?**
J. Butler; M. Main; M. Eagle; V. Decostre; V. Ricotti; F. Muntoni

G.P.396 **Mastication problems in Duchenne muscular dystrophy caused by dystrophic changes of the masseter muscle and abnormal dental characteristics**
L. van den Engel-Hoek; H. Hijdra; S. de Groot; L. Sie; C. Erasmus; I. de Groot

G.P.397 **Ankle-foot orthoses in Duchenne muscular dystrophy**
A. Mattiello-Sverzut; M. Souza; C. Baptista; M. Figueiredo; R. Alvades

G.P.398 **Kinesiophobia in Duchenne muscular dystrophy: perspective of parents and physiotherapists**
H. Alkan; N. Bulut; C. Bozdemir Özel; I. Öncel; O. Yılmaz; A. Karaduman; H. Topaloğlu

G.P.399 **The relation between visual-motor control and upper-limb speed and dexterity in Duchenne muscular dystrophy**
S. Karayazgan; S. Subası; N. Bulut; O. Yılmaz; A. Karaduman; B. Akel; H. Topaloğlu

G.P.400 **Implantable cardioverter defibrillator for cardiac resynchronisation therapy in Duchenne muscular dystrophy: A case study**
R. Kulshrestha; A. Morley-Davies; T. Willis

G.P.401 **Natural history of respiratory function changes in patients with Duchenne muscular dystrophy not using glucocorticoid steroids**
G. Buyse; T. Voit; U. Schara; C. Straathof; M. D'Angelo; G. Bernert; J. Cuisset; R. Finkel; N. Goemans; C. McDonald; C. Rummey; T. Meier

G.P.402 **Interaction between brain neuronal circuits of the face recognition memory area and the auditory memory area in Duchenne muscular dystrophy patients**
A. Secaf; J. Martins; M. Sawan; A. Godoy

G.P.403 **Dissociation between motor and cognitive skills in patients with Duchenne muscular dystrophy**
M. Voos; F. Favero; K. Dias; M. Artiheiro; A. Oliveira; F. Caromano

G.P.404 **The frequency and characterisation of cardiac involvement in female carriers of BMD or DMD: A cross sectional analysis**
T. McCaffrey; A. Murphy; M. Guglieri; K. Bushby; J. Bourke

Fasciitis and immune neuropathies (G.P.405–418)
Facilitators: Pieter van Doorn and Edoardo Malfatti

G.P.405 **A new method to determine muscle and movement action during daily activities in patients with neuralgic amyotrophy**
C. Cup; A. Bergsma; I. de Groot; J. Ijspeert; R. Janssen; J. Groothuis; N. van Alfen

G.P.406 **Neuromyotonia: Clinical, electrophysiological and immunological findings**
P. Lorenzoni; A. Miranda; L. Filla; C. Kay; A. Vicent; L. Werneck; R. Scola

G.P.407 **Acute myopathy as a manifestation of neuromyelitis optica**
H. So-Young; S. Kim; Y. Park; D. Kim; H. Kim; J. Kim; J. Shin

G.P.408 **Incidence of neuralgic amyotrophy (Parsonage–Turner syndrome) in a primary care setting: A prospective cohort study**
N. van Alfen; J. Groothuis; E. Nobacht; T. Ennik; S. Flynn; S. Pillen; J. van Eijk; F. van de Laar

G.P.409 **Small sensory fiber neuropathy in patients with long-lasting macrophagic myofasciitis**
J. Aouizerate; H. Sahli; Y. Baba Amer; J. Lefaucheur; F. Authier

G.P.410 **Neuralgic amyotrophy with phrenic neuropathy: A difficult diagnosis but not invariably a bad outcome**
N. van Alfen; J. Doorduin; Y. Heijdra; A. Boon; P. Wijkstra; J. Groothuis

G.P.411 **Two women with brachio-cervical inflammatory myopathy (BCIM) and fatal cardiomyopathy**
A. Van der Kooi; B. Jaeger; Y. Pinto; E. Aronica; M. de Visser

G.P.412 **Atypical Miller–Fisher syndrome presenting as acute isolated bilateral ophthalmoplegia in young children**
F. Guisset; C. Ferreiro; J. Selliera; F. Corazza; C. Prigogine; N. Deconinck

G.P.413 **Enhancement of myosin heavy chain class I (MHC I) mRNA expression in C2C12 myocyte by multivalent cations**
Y. Mori; J. Yamaji; R. Hiroshima; T. Nakano; M. Watanabe; A. Miyazaki

G.P.414 **Contribution of IL-6-dependent signalling mechanism to upregulation of MyHC IIb mRNA but not of MyHC IIa mRNA in mouse myocytes**
J. Yamaji; Y. Mori; R. Hiroshima; M. Watanabe; A. Miyazaki

G.P.415 **New targets to control skeletal muscle inflammation: MicroRNAs regulated by adiponectin**
R. Boursereau; M. Abou-Samra; S. Lecompte; C. Deprez; L. Noël; S. Brichard

G.P.416 **Perifascicular pathology in eosinophilic fasciitis with muscle involvement**
D. Pehl; C. Preusse; J. Rinnenthal; U. Schneider; F. Heppner; Y. Allenbach; O. Benveniste; H. Goebel; W. Stenzel

G.P.417 **Neuropsychological correlates of brain perfusion SPECT abnormalities in patients with Macrophagic myofasciitis**
A. Van der Gucht; M. Aoun Sebati; E. Itti; J. Aouizerate; A. Bachoud; F. Authier

G.P.418 **Wrist-worn accelerometer as innovative tool for longitudinal follow-up of idiopathic inflammatory myopathy patients: A pilot study**
Y. Allenbach; A. Foucher; N. Champtiaux; L. Gilardin; B. Hervier; O. Benveniste; J. Hogrel

17:30–19:30 **Symposium 3; BioMarin Pharmaceutical Inc.** – Concert Hall, including refreshments in Corn Exchange

Saturday 3 October 2014

08:00 **Conference desk opens** – Brighton Dome foyer

08:30–10:00 **Advances in treatment of neuromuscular disease;** Invited lectures (I.I.9–11) – Concert Hall
Chairpersons: Jenny Morgan and Kevin Flanigan

I.I.9 **Molecular regulation of muscle stem cell asymmetric division**
M. Rudnicki

I.I.10 **Signalling pathways and modifiers in cardiac function in muscular dystrophy**
E. McNally

I.I.11 **Gene delivery for treating neurological disorders**
B. Kaspar

10:00–10:30 **Morning refreshments, exhibition and posters** – Corn Exchange, café and mezzanine

10:30–12:00 **Pathophysiology of neuromuscular diseases;** Oral presentations (G.O.13–18) – Concert Hall
Chairpersons: Ichizo Nishino and Bjarne Udd

G.O.13 **A pre-clinical model of LGMD1D/DNAJB6 associated muscle disease: Expanding the role of HSP chaperones in skeletal muscle**
C. Weihl; S. Pittman; H. True; R. Bengochea

G.O.14 **Antisense strategies targeting DUX4 and DUX4c as therapeutic approaches for facioscapulohumeral muscular dystrophy (FSHD)**
C. Vanderplanck; A. Tassin; E. Ansseau; C. Lancelot; A. Derenne; S. Conotte; V. Dudome; B. Leroy; S. Wilton; D. Laoudj-Chenivesse; R. Wattiez; A. Legrand; A. Belayew; F. Coppée

G.O.15 **Correlation between low FAT1 expression and early affected muscle in FSHD**
V. Mariot; S. Roche; C. Hourdé; D. Portilho; S. Sacconi; F. Puppo; S. Duguez; P. Rameau; N. Caruso; A. Delezoide; C. Desnuelle; B. Bessières; S. Collardeau; L. Feasson; T. Maisonobe; F. Magdinier; F. Helmbacher; G. Butler-Browne; V. Mouly; J. Dumonceaux

G.O.16 **Differential isoform expression and selective muscle involvement in muscular dystrophies**
P. Hackman; S. Huovinen; S. Penttilä; J. Keto; P. Somervuo; P. Auvinen; A. Vihola; K. Pelin; O. Raheem; T. Suominen; B. Udd

G.O.17 **The physiological consequences of different levels of dystrophin following antisense based exon-skipping in the *mdx* mouse**
S. Muses; C. Godfrey; G. McClorey; K. Wells; T. Coursindel; R. Terry; C. Betts; O. Cappellari; S. Hammond; E. O'Donovan; J. Hildyard; S. El Andaloussi; M. Gait; M. Wood; D. Wells

G.O.18 **Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials**
E. Mercuri; R. Finkel; J. Montes; E. Mazzone; M. Main; D. Ramsey; A. Mayhew; A. Glanzman; A. Pasternak; M. Pane; M. Pera; M. Scoto; S. Messina; G. Vita; A. D'Amico; B. Darras; E. Bertini; F. Muntoni; D. De Vivo

12:15–13:45 **Symposium 4;** Genzyme Europe BV – Concert Hall

Lunch bags & afternoon excursion – Arundel Castle group visit

19:30–00:00 **Congress dinner** – Hilton Brighton Metropole

Sunday 4 October 2015

08:00 **Conference desk opens** – Brighton Dome foyer

08:30–10:30 **Advances in treatment of neuromuscular disorders;** Oral presentations (G.O.19–26) – Location: Concert Hall
Chairpersons: Ronald Cohn and Enrico Bertini

G.O.19 **Taurine supplemental therapy in prevention of stroke-like episodes in MELAS**
Y. Sunada; Y. Ohsawa; Y. Fukai; M. Fujino; S. Nishimatsu; S. Ohta

G.O.20 **Reveglucosidase alfa (BMN 701), a GILT-tagged recombinant human acid alpha glucosidase (rhGAA), evaluation in late-onset Pompe disease: Preliminary clinical efficacy and safety results of an extension study (72-week results)**
T. Hiwot; R. Barohn; D. Bratkovic; B. Byrne; C. Desnuelle; D. Hughes; P. Laforêt; E. Mengel; M. Roberts; K. Yang; C. Heusner; W. Dummer

G.O.21 **Reducing dynamin 2 rescues a severe congenital myopathy in mice**
B. Cowling; I. Prokic; T. Chevremont; C. Kretz; A. Ferry; C. Coirault; V. Laugel; N. Romero; J. Laporte

G.O.22 **Serum and urine proteomic profiling reveals biomarkers suitable for monitoring the outcome of therapeutic interventions in muscular dystrophies**
T. Voit; J. Poupiot; J. Camadro; C. Garcia; L. Servais; F. Svinartchouk; J. Rouillon; B. Wong; T. Léger; I. Richard

G.O.23 **Epicatechin enhances mitochondrial biogenesis, increases dystrophin and utrophin, increases follistatin while decreasing myostatin and improves skeletal muscle exercise response in adults with Becker muscular dystrophy (BMD)**
C. McDonald; E. Henricson; B. Oskarsson; C. Aguilar; A. Nicorici; N. Joyce; D. Reddy; A. Wagner; E. deBie; E. Goude; R. Abresch; F. Villareal; G. Perkins; Y. Hathout; S. Dugar; G. Schreiner

G.O.24 **Simvastatin improves physiological function and protects against muscle degeneration in *mdx* mice: A novel therapeutic approach for Duchenne muscular dystrophy**
N. Whitehead; M. Kim; K. Bible; M. Adams; S. Froehner

G.O.25 **Adeno-associated virus vector (AAV) microdystrophin gene therapy prolongs survival and restores muscle function in the canine model of Duchenne muscular dystrophy (DMD)**
C. Le Guiner; L. Servais; M. Montus; F. Bodvael; B. Gjata; J. Hogrel; P. Carlier; S. Moullec; C. Masurier; O. Adjali; F. Mingozzi; T. Koo; T. Athanasopoulos; Y. Cherel; F. Mavilio; T. Voit; P. Moullier; G. Dickson

G.O.26 **Development of LNA gapmer oligonucleotide based therapy for FTD/ALS caused by the C9orf72 repeat expansion**
Y. Aoki; M. Raquel; Y. Lee; A. Douglas; M. Aoki; M. Varela; C. Sathyaprakash; R. Mutihac; K. Talbot; M. Wood

10:30–11:00 **Morning refreshments, exhibition and posters** – Corn Exchange, café and mezzanine

11:00–12:30 **Poster highlights** – Concert Hall
Chairpersons: Thomas Voit and Ichizo Nishino

12:30–13:30 **WMS General Assembly** – Concert Hall

13:30–14:30 **Lunch, exhibition and posters** – Corn Exchange, café and mezzanine

14:30–16:30 **Late breaking session** – Concert Hall
Chairpersons: Norma Romero and Volker Straub

16:30 **Prize giving and welcome to the 21st WMS Congress.**
Handover of the WMS flag and close of Congress – Concert Hall