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Neuromuscular Disorders 27 (2017) S57–S94

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22nd WMS Congress – 2017 – Program

Tuesday 3 October 2017

12:30–16:00		**WMS Executive Board Meeting** – Hôtel France & Chateaubriand
14:00–18:00		**Registration opens** **Setting up of posters**
18:00–18:45		**Opening Ceremony –** Auditorium Chateaubriand
18:45–21:00		**Welcome Reception** – Grand Large room and Jacques Cartier rotunda

Wednesday 4 October 2017

08:00–19:30		**Conference desk opens**
08:30–09:00		**Congress welcome – Message from the President** – Auditorium Chateaubriand
09:00–10:30		**Excitation-contraction coupling: basic aspects & related disorders** Invited lectures (EC.I.1–3) *Chairpersons: Gisèle Bonne and Thomas Voit*
	EC.I.1	**Basic aspects of excitation-contraction coupling and its relation to clinical disorders** B. Allard
	EC.I.2	**Formation and maintenance of the neuromuscular junction: molecular mechanisms and physiological consequences** L. Schaeffer
	EC.I.3	**Clinical aspects of excitation-contraction coupling (ECC) disorders** H. Jungbluth
10:30–11:00		**Morning refreshments, exhibition and posters –** Grand Large room and Jacques Cartier rotunda
11:00–13:00		**Excitation-contraction coupling and muscle homeostasis** Oral presentations (EC.O.1–8) – Auditorium Chateaubriand *Chairpersons: Isabelle Marty and Helge Amthor*
	EC.O.1	**Genetic and histological characterisation of excitation-contraction coupling related structural myopathy cohort** S. Beecroft; R. Choi; C. McLean; M. Olivé; M. Ryan; M. Davis; N. Laing; B. Launikonis; G. Ravenscroft
	EC.O.2	**The blurred scenario of the new calcium-related myopathies: clinical, radiological and molecular characterization of *CASQ1*, *STIM1* and *ORAI1* myopathies diagnosed in Padova neuromuscular center** C. Semplicini; C. Bertolin; B. Pantic; L. Bello; S. Vianello; F. Catapano; I. Colombo; M. Moggio; G. Sorarù; G. Cenacchi; C. Calore; R. Stramare; E. Pegoraro
	EC.O.3	**Dysferlin reduces calcium leak and stabilizes excitation-contraction coupling in mature muscle** R. Bloch; V. Lukyanenko; J. Muriel

http://dx.doi.org/10.1016/j.nmd.2017.06.016

EC.O.4 **Reversible endogenous downregulation of myostatin pathway in wasting neuromuscular diseases explains challenges of anti-myostatin therapeutic approaches**
V. Mariot; R. Joubert; C. Hourdé; L. Féasson; M. Hanna; F. Muntoni; T. Maisonobe; L. Servais; R. Le Panse; O. Benveniste; T. Stojkovic; P. Machado; T. Voit; A. Buj-Bello; J. Dumonceaux

EC.O.5 **The multi-systemic protection against age-related tissue function decline in progeric mice through the attenuation of myostatin/activin signalling**
K. Patel; K. Alyodawi; S. Omairi; W. Vermeij; O. Kretz; F. Salagna; T. Huber

EC.O.6 **Abolition of the NLRP3 inflammasome improves the dystrophic phenotype in a murine model of Duchenne muscular dystrophy**
R. Boursereau; M. Abou-Samra; S. Lecompte; L. Noel; S. Brichard

EC.O.7 **Necroptosis, a programmed form of necrosis, participates in muscle degeneration in Duchenne muscular dystrophy**
M. Bencze; J. Meng; V. Pini; F. Conti; F. Muntoni; J. Morgan

EC.O.8 **Epigenetic regulation of a mitochondrial apoptosis mediator, harakiri in maintaining muscle membrane stability in autoimmune myositis**
K. Nagaraju; J. Boehler; A. Horn; J. Novak; S. Ghimbovschi; I. Lundberg; J. Jaiswal

13:00–14:30 **Lunch, exhibition and posters**

14:30–16:00 **Poster session 1: parallel sessions (P.1–134) –** Poster area

DMD general and clinical considerations (P.1–21)
Chairpersons: Nathalie Goemans and Basil Darras

P.1 **Clustering trajectories of ambulatory function in the North Star clinical network database**
F. Muntoni; J. Domingos; A. Manzur; A. Mayhew; M. Guglieri; J. Signorovitch; S. Ward

P.2 **The association between muscle weakness and gait deviations in children with Duchenne muscular dystrophy**
M. Goudriaan; N. Goemans; M. Van den Hauwe; K. Desloovere

P.3 **Has outcome changed for adults with Duchenne muscular dystrophy?**
N. Goemans; L. Dewaele; K. Claeys; M. Proesmans; B. Cools; M. vanden Hauwe; P. Moens; B. Vrijsen; B. Buyse

P.4 **The impact of an interdisciplinary DMD clinic model on volume of care recommendations**
L. McAdam; J. Setchell; P. Thille; T. Abrams; B. Mistry; B. Gibson

P.5 **DMD HUB: expanding clinical trial capacity for Duchenne muscular dystrophy in the UK**
E. Heslop; V. Straub; M. Guglieri; C. Turner; B. Davis; E. Crossley; A. Johnson; K. Bushby

P.6 **A natural history study of Becker muscular dystrophy by the CINRG investigators**
P. Clemens; N. Gonzalez; L. Morgenroth; A. Smith; G. Niizawa; J. Florence; K. Gorni; H. Abdel-Hamid; M. Guglieri; A. Connolly; T. Bertorini; M. Wicklund; E. Smith; N. Kuntz; M. Thangarajh; J. Mah; C. Spurney; C. McDonald; A. Cnaan; H. Gordish-Dressman; CINRG Investigators

P.7 **The relation of postural alignment and energy expenditure in boys with Duchenne muscular dystrophy**
S. Bozgeyik; I. Alemdaroğlu; A. Karaduman; H. Topaloğlu; O. Yılmaz

P.8 **Predictors of ambulation in patients with Duchenne muscular dystrophy**
E. Zapata Aldana; N. Eltayeb; M. Miller; C. Campbell

P.9 **The relation of respiratory and upper extremity functions in early stage of Duchenne muscular dystrophy: a pilot study**
N. Bulut; G. Aydın; A. Karaduman; I. Alemdaroğlu; H. Topaloğlu; O. Yılmaz

P.10 **Analysis of mortality in a cohort of adult Duchenne muscular dystrophy**
L. Nastase; M. Desikan; S. Price; F. Crummy; J. Kahn; R. Quinlivan

P.11 **Percutaneous endoscopic gastrostomy in patients with Duchenne muscular dystrophy compared with amyotrophic lateral sclerosis and Parkinson syndrome**
H. Arahata; T. Nishiyama; K. Inada; A. Miyoshi; A. Watanabe; Y. Kawano; N. Sasagasako; N. Fujii

P.12 **Acute effect of muscle facilitation application on performance and energy expenditure in Duchenne muscular dystrophy**
G. Aydın; I. Alemdaroğlu; A. Karaduman; H. Topaloğlu; O. Yılmaz

P.13 **Study on factors related to general condition and prognosis of patients with Duchenne muscular dystrophy**
T. Saito; K. Ogata; T. Takahashi; M. Kobayashi; T. Takada; S. Kuru; T. Mikata; T. Matsumura; T. Fukudome; M. Funato; H. Arahata; N. Yonemoto; E. Kimura

P.14 **Testosterone therapy in patients with Duchenne muscular dystrophy and glucocorticoid-induced pubertal delay**
C. Keefe; B. Wong; I. Rybalsky; K. Shellenbarger; C. Tian; J. Khoury; L. Hornung; M. Rutter

P.15 **Body composition of patients with Duchenne muscular dystrophy**
B. Wong; S. Hu; P. Horn; I. Rybalsky; K. Shellenbarger; C. Tian; J. Bange; H. Kalkwarf

P.16 **Relationships between ambulatory function and body composition in patients with Duchenne muscular dystrophy**
B. Wong; J. Signorovitch; S. Hu; J. Bange; I. Rybalsky; K. Shellenbarger; C. Tian; E. Swallow; J. Song; S. Ward

P.17 **Insights into bone mineral density and bone metabolism in Duchenne muscular dystrophy**
M. Sframeli; G. Vita; A. Catalano; M. Distefano; M. La Rosa; C. Barcellona; C. Bonanno; G. Nicocia; C. Profazio; N. Morabito; C. Lunetta; G. Vita; S. Messina

P.18 **The effects of calf massage in boys with Duchenne muscular dystrophy**
K. de Valle; E. Yiu; M. Ryan; A. Kornberg; R. Kennedy; D. Villano; K. Carroll

P.19 **Novel mutation in the dystrophin gene causing distal asymmetric muscle weakness of the upper limbs**
J. Afonso Ribeiro; L. Almendra; O. Rebelo; F. Laranjeiro; A. Marmiesse; M. Almeida; M. Peres; A. Geraldo; A. Matos; L. Negrao

P.20 **Comprehensive analysis: nonsense mutation induced exon skipping in Becker muscular dystrophy**
M. Okubo; S. Noguchi; E. Kimura; S. Mitsuhashi; I. Nishino

P.21 **Genotype-phenotype associations in a large cohort of Duchenne muscular dystrophy patients**
D. Moon; S. Hu; J. Bange; P. Horn; I. Rybalsky; K. Shellenbarger; B. Wong

Congenital muscular dystrophies: collagen VI and merosin (P.22–34)
Chairpersons: Valérie Allamand and Carsten Bönnemann

P.22 **Clinical and genetic characterization of collagen VI-related myopathies: difficulties in phenotypic characterization in the first years of life**
D. Natera-de Benito; M. Alarcon; C. Ortez; A. Nascimento; C. Jou; J. Medina; M. Vigo; A. Codina; A. Frongia; J. Colomer; C. Jiménez-Mallebrera

P.23 **Bethlem myopathy, the other side of collagen VI myopathies**
A. Martins; M. Almeida; A. Geraldo; A. Matos; L. Negrao

P.24 **A possible new phenotype associated with variants in *COL6A2* gene**
S. Lehtinen; S. Penttilä; T. Suominen; A. Väisänen; J. Weinberg; G. Solders; M. Arumilli; P. Hackman; B. Udd

P.25 **A common *COL6A1* deep-intronic pseudo-exon inserting mutation causes a distinct phenotype of Ullrich congenital muscular dystrophy**
A. Reghan Foley; S. Donkervoort; V. Bolduc; Y. Hu; B. Cummings; M. Lek; A. Sarkozy; C. Jiménez-Mallebrera; R. Butterfield; S. Lamande; J. Kirschner; V. Allamand; T. Stojkovic; S. Quijano-Roy; F. Gualandi; A. Ferlini; E. Bertini; D. MacArthur; F. Muntoni; C. Bönnemann

P.26 ***COL6A* genes transcriptomic by RNAseq and fluidic card tools**
R. Rossi; C. Scotton; M. Falzarano; A. Armaroli; H. Osman; M. Neri; G. Pesole; M. Mora; E. Pegoraro; L. Merlini; A. Ferlini; F. Gualandi

P.27 **Collagen VI deficiency: the heart of the matter**
M. Saunier; C. Gartioux; M. Beuvin; N. Mougenot; G. Bonne; V. Allamand

P.28 **A mouse with exon 9 deletion in *Col6a1* as a model for dominant collagen VI-related disorders**
S. Noguchi; M. Ogawa; I. Nishino

P.29 **Gapmer antisense oligonucleotides selectively suppress the mutant allele of *COL6A3* gene in dominant Ullrich congenital muscular dystrophy**
H. Zhou; E. Marrosu; P. Ala; F. Muntoni

P.30 **Congenital muscular dystrophy ascending multiple dose cohort study analyzing pharmacokinetics at three dose levels in children and adolescents with assessment of safety and tolerability of omigapil (CALLISTO) trial update**
M. Leach; A. Foley; G. Averion; Y. Hu; P. Yun; J. DeCoster; C. Arevalo; C. Mendoza; O. Mayer; R. Hausmann; K. Cheung; C. Bönnemann

P.31 **A diagnostic challenge of late onset adult merosinopathy**
C. von Landenberg; K. Kappes-Horn; M. Stepien-Mering; J. Reimann

P.32 **Electrophysiological and pathological studies of peripheral nerves in children with merosin-deficient congenital muscular dystrophy type 1A**
Y. Saito; A. Ishiyama; Y. Saito; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; K. Sugai; I. Nishino; M. Sasaki

P.33 **Jab1 in the pathogenesis of merosin - deficient congenital muscular dystrophy (MDC1A)**
D. Velardo; E. Porrello; R. Tonlorenzi; I. Lorenzetti; R. Pardi; D. Goldhamer; S. Previtali

P.34 **Limb-girdle muscular dystrophy related to *LAMA2* mutations: an unusual familial coincidence responsible for the phenotypic variability and diagnostic difficulties**
V. Guillet-Pichon; F. Leturcq; K. Claeys; C. Beroud; A. Nadaj-Pakleza

Congenital muscular dystrophies: dystroglycanopathies and others (P.35–48)
Chairpersons: Rita Barresi and Adele D'Amico

P.35 **Translational biochemistry for dystroglycanopathies**
H. Wang; R. Sprute; H. Daimagüler; S. Cirak

P.36 **Limb girdle muscular dystrophy type 2I: lack of correlation between clinical severity, histopathological alterations and levels of glycosylated α-dystroglycan in patients homozygous for the common *FKRP* mutation**
S. Lindal; M. Alhamidi; V. Brox; E. Stensland; M. Liset; O. Nilssen

P.37 **Cardiac involvement in a patient with congenital muscular dystrophy related to *POMT2* gene mutation**
M. Sframeli; M. La Rosa; M. Distefano; C. Barcellona; G. Vita; G. Nicocia; G. Astrea; A. D'Amico; E. Bertini; F. Santorelli; G. Vita; S. Messina

P.38 **The phenotype of *POGLUT1* mutations: broad clinical expression and distinctive muscle imaging pattern**
E. Servián-Morilla; M. Cabrera-Serrano; H. Takeuchi; N. Muelas; E. Rivas-Infante; G. Cantero; F. Mavillard; J. Vilchez; C. Paradas

P.39 **Autosomal recessive myopathy associated with cataracts and learning difficulties caused by *INPP5K* mutations: a new syndromic gene linking four overlapping rare recessive neuromuscular disorders**
A. Roos; J. Senderek; D. Cox; M. Wiessner; R. Zahedi; R. Charlton; R. Barresi; D. Hathazi; H. Lochmüller

P.40 **Congenital muscular dystrophy with cataracts and mild cognitive impairment cause by mutations in *INPP5K*: overlapping Marinesco-Sjögren syndrome and dystroglycanopathy**
A. Nascimento; C. Ortez; C. Jou; M. Alarcón; A. Bates; A. Topf; D. Itzep; D. Natera; A. Frongia; A. Codina; L. Gonzalez; G. Pia; V. Straub; C. Jiménez-Mallebrera; J. Colomer

P.41 **A novel *INPP5K* mutation in a sibship from the Reunion Island**
I. Nelson; M. Jacquemont; A. Urtizberea; M. Renouil; A. Boland; C. Masson; R. Ben Yaou; G. Bonne

P.42 **Congenital muscular dystrophy in Taiwan: a referral center experience**
W. Liang; X. Tian; C. Yuo; W. Chen; T. Kan; Y. Su; I. Nishino; L. Wong; Y. Jong

P.43 **A rare form of congenital muscle disorders; megaconial congenital muscular dystrophy**
A. Aksoy; Ö. Köken; H. Özyürek; B. Talim

P.44 **A case of severe encephalopathy and movement disorder due to mutations in the *TRAPPC11* gene**
A. Nascimento; C. Ortez; J. Colomer; D. Natera; A. Frongia; M. Alarcon; D. Itzep; C. Jou; A. Codina; J. Corbera; M. Rodriguez; M. Rodriguez; L. González; P. Gallano; M. Sacher; A. Topf; V. Straub; C. Jiménez-Mallebrera

P.45 **Complications of advanced Fukuyama congenital muscular dystrophy from a nationwide registry**
K. Ishigaki; C. Ihara; T. Sato; M. Shichiji; T. Murakami; K. Ishiguro; M. Mori-Yoshimura; H. Kaiya; M. Osawa; S. Nagata

P.46 **A case of epidermolysis bullosa simplex and muscular dystrophy with myasthenic symptoms caused by two novel *PLEC* mutations**
J. Lee; H. Shin; Y. Choi

P.47 **Congenital centronuclear myopathy and epidermolysis bullosa due to two novel mutations in the plectin gene**
M. Walter; P. Reilich; S. Krause; A. Abicht; B. Schoser

P.48 **Novel compound heterozygous mutations in *PLEC* gene causing epidermolysis bullosa simplex with muscular dystrophy, case series of two affected sisters**
I. Lee; A. Hurst; B. Wong; C. Tian

DMD/BMD: heart, brain and lungs (P.49–62)
Chairpersons: Brenda Wong and Craig McDonald

P.49 **Duchenne muscular dystrophy patients with chest pain require cardiac evaluation: a report of eight DMD patients presenting with chest pain, marked troponin elevation, and worsening cardiomyopathy**
P. Johnston; L. Cripe; K. Hor; C. Stiver

P.50 **Adolescence with Duchenne and Becker muscular dystrophy: a cardiac magnetic resonance comparison study**
P. Johnston; K. Hor; L. Cripe

P.51 **Quantitative cardiac NMR imaging in a large cohort of patients with Becker muscular dystrophy**
B. Marty; M. Toussaint; R. Gilles; P. Carlier; K. Wahbi

P.52 **Long-term effects of TRPV2 inhibition therapy for cardiomyopathy of muscular dystrophy**
T. Matsumura; M. Matsui; Y. Iwata; M. Asakura; T. Saito; H. Fujimura; S. Sakoda

P.53 **Effects of long-term treatment with eteplirsen on cardiac function**
L. Cripe; S. Colan; H. Eliopoulos; S. Moody; J. Mendell

P.54 **Increased blood pressure and BMI in relation to cardiomyopathy in Duchenne muscular dystrophy**
N. van de Velde; E. van Zwet; A. Roest; E. Niks

P.55 **Brain involvement in Duchenne muscular dystrophy: a role for dystrophin isoform Dp71 in cell migration and proliferation**
A. Ash; L. Booth-Wynne; K. Anthony

P.56 **Predominant posterior cerebral cortical atrophy in patients with *DMD* mutations**
J. Domingues; P. Tavares; L. Souza; T. Rezende; R. Casseb; A. Martinez; L. Rittner; S. Appenzeller; A. Nucci; M. França Junior

P.57 **Timing and localization of human dystrophin isoform expression provide insights into the cognitive phenotype of Duchenne muscular dystrophy**
N. Doorenweerd; A. Mahfouz; M. van Putten; R. Kaliyaperumal; P. 't Hoen; J. Hendriksen; A. Aartsma-Rus; J. Verschuuren; E. Niks; M. Reinders; H. Kan; B. Lelieveldt

P.58 **Longitudinal pulmonary function testing outcome measures in Duchenne muscular dystrophy: long-term natural history with and without glucocorticoids**
C. McDonald; H. Gordish-Dressman; E. Henricson; T. Duong; N. Joyce; S. Jhawar; M. Leinonen; F. Hu; A. Connolly; A. Cnaan; R. Abresch; CINRG Investigators

P.59 **Long-term benefits of glucocorticoids in Duchenne muscular dystrophy: is it worth it?**
C. McDonald; E. Henricson; R. Abresch; T. Duong; N. Joyce; F. Hu; P. Clemens; E. Hoffman; A. Cnaan; H. Gordish-Dressman; CINRG Investigators

P.60 **Co-morbidities in a cohort of adult Duchenne muscular dystrophy patients attending a Neuromuscular Complex Care Centre – an observational study**
M. Desikan; L. Nastasi; S. Price; R. Scalco; J. Pattni; M. Hanna; R. Quinlivan

P.61 **Myoglobinuria in two patients with Duchenne muscular dystrophy after treatment with zoledronate: a case-report and call for caution**
C. Bloetzer; D. Jacquier; A. Klein; A. Ivanyuk; N. Garcia Segarra

P.62 **Genotype-phenotype correlations in a French cohort with Becker muscular dystrophy: focus on cognitive aspects**
F. Dahimene; A. Nadej-Pakleza; J. Durigneux; S. Mercier; Y. Pereon; A. Magot

Mitochondrial disease and myofibrillar myopathies (P.63–80)
Chairpersons: Montse Olivé and Pascal Laforêt

P.63 **TIEG1 is a novel regulator of muscle mitochondrial biogenesis**
M. Kammoun; V. Veksler; J. Piquereau; G. Bonne; M. Beuvin; I. Nelson; P. Pouletaut; M. Subramaniam; J. Hawse; S. Bensamoun

P.64 **Insights into the genesis of a brain and muscle disorder caused by a novel mutation in *MICU1***
A. Roos; A. Topf; V. Phan; J. Gonzalez Coraspe; M. Hauessler; J. Weis; H. Lochmüller; N. Kohlschmidt

P.65 **Profound hypotonia, muscle weakness, global developmental delays with stepwise regression, and cerebellar atrophy: expansion of the *LONP1*-related disease phenotype**
G. Nimmo; A. Pandey; C. Marshall; S. Venkatesh; L. Hazrati; S. Ahmed; J. Cameron; P. Ray; C. Suzuki; G. Yoon

P.66 **Acylcarnitine profile mimicking multiple acyl-CoA dehydrogenase deficiency in a patient with mitochondrial myopathy and a mutation in *MT-CO2***
S. Roos; K. Sofou; C. Hedberg-Oldfors; G. Kollberg; U. Lindgren; C. Thomsen; J. Asin-Cayuela; M. Tulinius; A. Oldfors

P.67 **Integrated genome analysis of COX deficiency in Japan**
K. Takayama; A. Iida; S. Noguchi; I. Nonaka; Y. Goto; I. Nishino

P.68 **Correlation of serum biomarkers and magnetic resonance spectroscopy (MRS) in monitoring disease progression in patients with mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) due to mtDNA A3243G mutation**
H. Lee; Y. Lee

P.69 **The evaluation of new biomarkers in a case of MELAS**
M. Matsui; M. Yamadera; T. Saito; T. Matsumura; H. Fujimura

P.70 **Mitochondrial disorders in adults: clinical and genetic correlations in ten patients with nuclear DNA mutations**
B. Kierdaszuk; M. Kaliszewska; K. Tonska; E. Bartnik; A. Kaminska; A. Kostera-Pruszczyk

P.71 **Exercise intolerance in a large multigeneration family associated with a homoplasmic mitochondrial DNA mutation in *MT-TL1***
N. Darin; C. Hedberg-Oldfors; A. Kroksmark; A. Moslemi; G. Kollberg; A. Oldfors

P.72 **GDF-15 expression in mitochondrial and other neurological diseases**
A. Ishii; S. Yatsuga; Y. Koga; A. Tamaoka

P.73 **Comprehensive genome analysis of Japanese patients with myofibrillar myopathy**
M. Inoue; A. Iida; S. Noguchi; I. Nonaka; I. Nishino

P.74 **A novel mutation in *HSPB8* causes dominant adult-onset axial and distal myopathy**
A. Echaniz-Laguna; X. Lornage; B. Lannes; R. Schneider; G. Bierry; N. Dondaine; J. Deleuze; J. Böhm; J. Thompson; J. Laporte; V. Biancalana

P.75 **A novel *BAG3* mutation associated with myofibrillar myopathy emphasizes HSP70 dependent activity**
R. Bengoechea; W. Motley; S. Bird; S. Zuchner; S. Scherer; C. Weihl

P.76 **Reducing body myopathy as a new phenotype of Filamin C mutation**
A. Janin; V. Manel; G. Millat; N. Streichenberger

P.77 **Increased cardiac muscle autophagy in a child with restrictive cardiomyopathy, proximal myopathy and neuropathy due to a mutation in the *BAG3* (Pro209Leu) gene**
A. Schänzer; S. Rupp; G. Mall; H. Akintürk; D. Schramz; L. Gulatz; J. Thul; N. Mazhari; A. Hahn

P.78 **Expression of myofibrillar myopathy gene products in the nervous system**
Y. Blech-Hermoni; S. Coscia; L. Jensen; M. Kates; K. Subedi; A. Mankodi

P.79 **Mouse model of *BAG3* myofibrillar myopathy**
R. Robertson; T. Conte; M. Dicaire; R. Bryson-Richardson; J. Lavoie; E. O'Ferrall; J. Young; B. Brais

P.80 **Identification of therapies for myofibrillar myopathy**
A. Ruparelia; C. Williams; E. McKaige; V. Oorschot; E. Baxter; K. Schulze; G. Ramm; R. Bryson-Richardson

MRI in neuromuscular diseases and new techniques (P.81–100)
Chairpersons: Pierre Carlier and Hermien Kan

P.81 **Adding quantitative muscle MRI to the FSHD clinical trial toolbox**
K. Mul; S. Vincenten; N. Voermans; S. van der Maarel; G. Padberg; C. Horlings; B. van Engelen

P.82 **Long-term follow-up of MRI changes in thigh muscles of patients with facioscapulohumeral dystrophy: a quantitative study**
E. Salort-Campana; F. Fatehi; A. Le Troter; E. Lareau-Trudel; M. Bydder; M. Guye; D. Bendahan; S. Attarian

P.83 **Muscle MRI findings in spinal muscular atrophy type 3**
H. Karasoy; T. Ozkan; M. Argin; A. Yuceyar; O. Ekmekci

P.84 **Magnetic resonance image in oculopharyngeal muscular dystrophy**
A. Alonso-Jimenez; A. Alejaldre-Monforte; C. Dominguez-Gonzalez; E. Cortes-Vicente; R. Rojas-Garcia; G. Tasca; R. Carlier; M. Monforte; P. Laforêt; G. Gutierrez-Gutierrez; A. Lopez de Munain; R. Fernandez-Torron; I. Illa; J. Diaz-Manera

P.85 **Usefulness of MRI in cases of hyperCKemia**
P. Marti; N. Muelas; J. Diaz-Manera; J. Vilchez

P.86 **Muscle alterations in sporadic inclusion body myositis assessed using quantitative nuclear magnetic resonance imaging and spectroscopy, ultrasound shear-wave elastography, and relationships with muscle function**
D. Bachasson; H. Reyngoudt; S. Turk; O. Benveniste; J-Y. Hogrel; P. Carlier

P.87 **Muscle magnetic resonance imaging in inclusion body myositis: presentation of 16 cases**
M. Rugiero; M. Bettini; M. Araoz; N. Genco; M. Chaves; S. Christiansen; A. Rassumoff

P.88 **Muscle MRI protocol for progression evaluation in inclusion body myositis and Becker muscular dystrophy-baseline data**
L. Maggi; M. Pasanisi; F. Mazzi; M. Verri; R. Frangiamore; M. Moscatelli; L. Chiapparini; R. Mantegazza; M. Bruzzone; D. Aquino

P.89 **^{31}P and ^{1}H nuclear magnetic resonance spectroscopy characterization of skeletal muscle pH dysregulation in Duchenne muscular dystrophy patients at rest**
H. Reyngoudt; S. Turk; P. Carlier

P.90 **Nuclear magnetic resonance relaxometry characterization of D2-mdx mice**
A. Martins-Bach; E. Araujo; B. Matot; Y. Fromes; P. Baudin; I. Richard; P. Carlier

P.91 **Quantitative analysis of muscle resonance imaging of fatty infiltration of the pelvic and lower limb muscles in Duchenne muscular dystrophy using excel based auto calculation program**
A. Hirasawa; A. Ishiyama; T. Nakayama; S. Kuru; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; I. Nishino; M. Sasaki

P.92 **Magnetic resonance biomarkers in the proximal and distal upper extremity in a large cohort of boys with Duchenne muscular dystrophy**
R. Willcocks; S. Forbes; D. Lott; C. Senesac; H. Arora; A. Barnard; A. Harrington; M. Daniels; E. Finanger; G. Tennekoon; R. Finkel; D. Wang; W. Rooney; G. Walter; H. Sweeney; K. Vandenborne

P.93 **Spatially localized phosphorous metabolism of skeletal muscle in Duchenne muscular dystrophy patients: 24-month follow-up**
M. Hooijmans; N. Doorenweerd; C. Baligand; J. Verschuuren; I. Ronen; A. Webb; E. Niks; H. Kan

P.94 **Stimulated echo DTI of skeletal muscle in Becker muscular dystrophy: a pilot study**
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P.95 **Skeletal muscle tissue characterization of a large cohort of patients with Becker muscular dystrophy using quantitative NMR imaging**
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P.96 **Effects of water compartmentation and distribution on skeletal muscle T1 values assessed by quantitative NMR imaging**
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P.100 **Auto calculation of muscle impairment ratio utilizing Mercuri grades from CT and MR images of muscle**
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P.102 **Registry of congenital neuromuscular disorders in Japan: establishment and implementation**
A. Ishiyama; E. Kimura; H. Nakamura; H. Komaki; M. Sasaki; I. Nishino

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S. Cameron; G. Eglon; L. Hastings; V. Straub; H. Lochmüller; K. Bushby; M. Guglieri; C. Marini-Bettolo

P.116 **Understanding decision needs for respiratory interventions in paediatric neuromuscular disorders**
D. Love; G. Mabaya; S. Katz; M. Lawson; A. Price; D. Radhakrishnan; J. Mah; L. Korngut; H. McMillan; C. Scholtes; A. Shephard; L. Hoey; M. Heletea; C. Campbell

P.117 **Men with Duchenne muscular dystrophy and end of life planning**
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P.118 **Prospective cohort study of spinal muscular atrophy types 2 and 3 in Spanish population**
D. Natera-de Benito; M. Alarcon; A. Borrás; J. Armas; A. Frongia; D. Itzep; M. Vigo; J. Medina; C. Ortez; J. Colomer; A. Nascimento

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M. Deguise; Y. De Repentigny; A. Beauvais; M. Bowerman; R. Kothary

P.120 **Segmental body composition in young children with SMA type 2: correlation with motor function abilities**
G. Baranello; M. Arnoldi; R. Zanin; R. Masson; C. Mastella; R. De Amicis; A. Battezzati; S. Bertoli

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P.122 **Autonomic nervous system involvement in spinal muscular atrophy type 1, 2 and 3**
S. Messina; M. Sframeli; G. Vita; C. Stancanelli; C. Terranova; E. Rizzo; F. Cavallaro; P. Girlanda; G. Vita

P.123 **Scoliosis is an inescapable comorbidity in SMA type II. A single center experience**
M. Catteruccia; G. Colia; A. Bonetti; A. Carlesi; L. Oggiano; G. La Rosa; F. Turturro; E. Bertini; E. Bertini; A. D'Amico

P.124 **Longitudinal data of the European prospective natural history study of patients with type 2 and 3 spinal muscular atrophy**
A. Chabanon; M. Annoussamy; A. Daron; Y. Pereon; C. Cancès; C. Vuillerot; N. Goemans; J. Cuisset; V. Laugel; U. Schara; E. Gargaun; T. Gidaro; A. Seferian; L. Lowes; P. Carlier; J-Y. Hogrel; C. Czech; R. Hermosilla; O. Khwaja; L. Servais

P.125 **Palliative care in children with spinal muscular atrophy type 1: how do they die? Results from a French multicentric study (National Hospital Clinical Research Program)**
M. Hully; C. Barnerias; S. Vanesse; M. Viallard; I. Desguerre

P.126 **Spinal muscular atrophy with lower extremity predominance: a recognizable phenotype of *BICD2* mutations in children**
R. Kulshrestha; C. Sewry; F. Muntoni; T. Willis; P. Munot

P.127 **Phenotypic heterogeneity in patients with mutations in the *IGHMBP2* gene**
M. Gomez-Garcia de la Banda; A. Felipe-Rucian; D. Gomez; M. Gratacos; A. Sanchez-Montañez; F. Gran; S. Bernal; E. Tizzano; J. Gamez; F. Munell

P.128 **Broad clinical spectrum observed in patients with scapuloperoneal spinal muscular atrophy (SPSMA) caused by an c.806G>A (p.Arg269His) mutation in the *TRPV4* gene**
M. Jędrzejowska; E. Dębek; P. Halat; A. Kostera-Pruszczyk; A. Jezela-Stanek; E. Ciara; M. Rydzanicz; P. Gasperowicz; M. Gos

P.129 **The recessive *SOD1* mutation p.A90V may cause atypical motor neuron disease by oligogenic mechanisms**
M. Jokela; S. Penttilä; B. Udd

P.130 **Differential expression of microRNAs in spinal muscular atrophy points towards significant peripheral organ involvement in the disease**
H. Zhou; M. Scoto; F. Catapano; I. Zahariewa; F. Muntoni

P.131 **Role of muscle satellite cells in spinal muscular atrophy physiopathology**
J. Mecca; S. Astord; T. Marais; F. Relaix; N. Didier; M. Barkats

P.132 **AAV9-mediated SMN expression restricted to the CNS does not rescue SMA mice**
A. Besse; M. Roda; S. Astord; T. Marais; M. Biferi; M. Barkats

P.133 **Identification of variants that affect severity of the spinal muscular atrophy phenotype within and outside of the *SMN2* gene**
A. Burghes; V. McGovern; C. Ruhno; T. Prior; P. Snyder; J. Roggenbuck; V. Sansone; J. Kissel

P.133B **The impact of the national population carrier screening program on reducing birth rates of patients with spinal muscular atrophy**
S. Aharoni; Y. Nevo; N. Orenstein; L. Basel-Vanagaite; H. Mussaffi; A. Singer

15:45–16:15 **Afternoon refreshments, exhibition and posters –** Grand Large room and Jacques Cartier rotunda

16:00–17:30 **Poster session 2: parallel sessions (P.134–264) –** Poster area

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P.134 **Clinical and molecular spectrum of early onset *LMNA*-related muscular dystrophy in Korea**
S. Choi; A. Cho; S. Kim; B. Lim; H. Kim; H. Hwang; K. Kim; J. Chae

P.135 **First results from the international *LMNA*-related congenital and childhood onset muscular dystrophy retrospective natural history study**
R. Ben Yaou; I. Dabaj; P. Yun; G. Norato; H. Xiong; A. Nascimento; L. Maggi; A. Sarkozy; S. Monges; M. Bertoli; H. Komaki; E. Mercuri; E. Zanoteli; K. Bushby; F. Muntoni; A. Rutkowski; C. Bönnemann; S. Quijano-Roy; G. Bonne; LMNA-rNHS Study Group

P.136 **Random forest approach to assess relationships of subjective muscle fatty infiltration with age at clinical onset and time of disease evolution in *LMNA*-related muscle disorders**
D. Gomez Andres; J. Diaz Manera; M. Gómez Garcia de la Banda; A. Sánchez-Carpintero; A. Alonso-Jimenez; A. Alejaldre-Monforte; F. Munell; R. Carlier; S. Quijano-Roy

P.137 **Corticosteroid treatment in early-onset lamin A/C related muscular dystrophies**
I. Dabaj; R. Ben Yaou; C. Bönnemann; A. Nascimento; A. Rutkowski; R. Erazo Torricelli; F. Muntoni; E. Lagrue; J. Dowling; K. Bushby; C. Casteglioni; K. Kleinsteuber; M. Lorenzo; A. Ishiyama; T. Sejersen; J. Gurgel-Giannetti; S. Monges; G. Bonne; S. Quijano-Roy

P.138 **Two novel mutations in the *FHL1* gene extending the phenotypic spectrum**
E. Strehle; K. Johnson; V. Rakocevic-Stojanovic; S. Peric; M. Farrugia; C. Longman; V. Straub

P.139 **A novel mutation in *FHL1* gene causing hypertrophic cardiomyopathy associated with myopathy**
R. Petillo; P. D'Ambrosio; C. De Luca; A. Papa; M. Iascone; G. Melloni; M. Scutifero; A. Palladino; L. Politano

P.140 **Clinical and histopathological characterization of the first French case of *MATR3*-related distal myopathy**
P. Laforêt; E. Malfatti; C. Metay; V. Jobic; R. Carlier

P.141 **Abnormal trafficking of connexin 43: a key element in the development of *LMNA* cardiomyopathy**
C. Macquart; M. Chatzifrangkeskou; M. Gotthardt; G. Bonne; A. Muchir

P.142 **Novel recessive splice site mutation in *POPDC1* (*BVES*) is associated with first-degree atrioventricular block and muscular dystrophy**
I. Nelson; M. Beuvin; R. BenYaou; C. Masson; A. Boland; R. Schindler; T. Brand; B. Eymard; G. Bonne

P.143 ***POPDC1* gene mutation screening in patients with LGMD and heart disturbances: a mutation load effect?**
R. Rossi; C. Scotton; X. Clinical Study group; P. Barton; R. Buchan; R. Walsh; S. Cook; H. Milting; G. Bonne; T. Brand; A. Ferlini

P.144 **Turnover studies on DNAJB6 and the CASA pathway proteins**
J. Sarparanta; S. Kawan; P. Jonson; B. Udd

P.145 **Molecular pathogenesis of caveolin-3-related limb-girdle muscular dystrophy**
J. Gonzalez Coraspe; D. Hathazi; H. Lochmüller; S. Carr; Y. Sunada; J. Weis; A. Roos

P.146 **A large multi-generation family with a novel *CAV3* mutation highlights extreme phenotypic variability and early childhood presentations**
C. Longman; M. Farrugia; A. Topf; D. Hathazi; R. Barresi; W. Stewart; A. Roos; I. Horrocks

P.147 ***CAV3* p.Ala93Thr pathogenic mutation causing hypertrophic cardiomyopathy**
R. Scalco; K. Savvatis; M. Desikan; M. Parton; C. Turner

P.148 **A case of caveolinopathy presenting as mounding muscle disease**
S. Chae; S. Kim; D. Kim; J. Shin

P.149 **Characteristic findings of skeletal muscle MRI in childhood-onset rippling muscle disease**
K. Ishiguro; T. Murakami; S. Kajino; M. Shichiji; T. Sato; Y. Hayashi; T. Nakayama; S. Kuru; M. Osawa; S. Nagata; K. Ishigaki

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Chairpersons: Isabelle Richard and Guillaume Bassez

P.150 **Clinical-molecular approach in autosomal recessive limb girdle muscular dystrophy**
R. Escobar Cedillo; M. Curiel Leal; A. Miranda Duarte; L. Sanchez Chapul; A. Luna Angulo; A. Cedeño; A.J. Urtizberea; M. Krahn; I. Viéitez González; C. Navarro Fernandez; B. Gomez; B. Vargas

P.151 **Limb girdle muscular dystrophy 2G in a religious minority of Bulgarian muslims homozygous for the c.75G>A, p.Trp25* *TCAP* mutation**
T. Chamova; S. Bichev; M. Gospodinova; D. Zlatareva; A. Taneva; K. Kastreva; T. Todorov; A. Todorova; I. Tournev

P.152 **Detection of *TRIM32* variants associated with LGMD2H in a large cohort of patients with unexplained limb-girdle weakness**
K. Johnson; A. Töpf; M. Bertoli; L. Phillips; W. De Ridder; J. Baets; P. De Jonghe; T. Deconinck; V. Rakocevic Stojanovic; S. Perić; H. Durmus; S. Jamal-Omidi; S. Nafissi; A. Łusakowska; T. Mongini; M. Lek; E. Valkanas; T. Mullen; L. Xu; D. MacArthur; V. Straub

P.153 **A new actinopathy – adult onset LGMD**
J. Palmio; P. Jonson; M. Savarese; S. Penttilä; S. Huovinen; M. Lindfors; B. Udd

P.154 **Severe respiratory involvement in a young-onset limb-girdle muscular dystrophy 2A from Guinea-Bissau associated with a novel pathogenic mutation**
M. Oliveira Santos; R. Roque; I. Conceição

P.155 **Characterization of the eosinophilic myositis caused by *CAPN3* mutations on a mouse model**
J. Warnez-Soulie; B. Giannesini; S. Henri; I. Richard; B. Malissen; M. Krahn; M. Bartoli

P.156 **Preclinical studies for calpain 3 gene transfer: primate pilot study and dose effect evaluation**
W. Lostal; C. Roudaut; V. Guilloux; S. Moullec; T. Larcher; J. Deschamps; I. Richard

P.157 **Calpain-3 stability following delays in freezing skeletal muscle biopsy samples – establishing an optimal time frame for accurate interpretation**
P. Kennedy; C. McLean; G. Lamb; R. Murphy

P.158 **Anoctamin 5 muscular dystrophy mimicking metabolic myopathy**
H. Durmus; R. Scalco; A. Gardiner; A. Manole; A. Schapira; J. Morrow; H. Houlden; J. Holton; K. Johnson; A. Töpf; L. Phillips; F. Deymeer; Y. Parman; V. Straub; R. Quinlivan; P. Oflazer-Serdaoglu

P.159 **The 107 kD *ANO5* protein is decreased in anoctaminopathy patients**
A. Vihola; H. Luque; M. Savarese; S. Penttilä; M. Lindfors; F. Leturcq; B. Eymard; G. Tasca; B. Brais; T. Conte; K. Charton; I. Richard; B. Udd

P.160 **Is cardiac dysfunction a feature of dysferlinopathy? Data from the clinical outcome study of dysferlinopathy**
R. Fernandez Torron; E. Harris; J. Bourke; K. Bettinson; H. Hilsden; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; J. Diaz-Manera; E. Pegoraro; J. Mendell; K. Bushby; V. Straub

P.161 **North Star Assessment for dysferlinopathy: longitudinal performance in the clinical outcome study of dysferlinopathy**
M. James; A. Mayhew; M. Eagle; R. Muni Lofra; E. Maron; R. Gee; M. Harman; T. Duong; B. Vandevelde; C. Siener; S. Thiele; J. Mendez; A. Canal; C. Sakamoto; S. Holsten; I. Pedrosa-Hernández; C. Semplicini; L. Lowes; K. Bushby; V. Straub

P.162 **Does the performance of upper limb capture functional variations in dysferlinopathy?**
M. James; A. Mayhew; D. Moat; M. Eagle; E. Maron; R. Gee; K. Rose; B. Drogo; B. Vandevelde; K. Foy; S. Thiele; M. Sánchez-Aguilera Práxedes; A. Canal; H. Yajima; M. Sanjak; E. Montiel-Morillo; L. Bello; L. Alfano; K. Bushby; V. Straub

P.163 **Examining the relationship between Dixon quantitative MRI and physiotherapy functional outcome measures in dysferlinopathy**
R. Fernandez Torron; M. James; F. Smith; N. Azzabou; I. Wilson; H. Reyngoudt; A. Mayhew; A. Blamire; P. Carlier; H. Hilsden; L. Rufibach; M. Jacobs; K. Bushby; V. Straub

P.164 **Novel binding partner of dysferlin is required for plasma-membrane repair**
H. Ono; N. Suzuki; S. Kanno; R. Izumi; T. Takahashi; Y. Kitajima; S. Osana; T. Akiyama; K. Ikeda; T. Shijo; S. Mitsuzawa; H. Warita; R. Nagatomi; N. Araki; A. Yasui; K. Miyake; M. Aoki

P.165 **New insights into lipid accumulation in dysferlin deficient muscular dystrophies**
M. Grounds; R. White; E. Lloyd; G. Pinniger

P.166 **A knock-in mouse model with nonsense dysferlin mutation**
J. Shin; S. Park; J. Park; D. Kim

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P.167 **Severe form of recessive Charcot-Marie-Tooth disease with a novel mutation in myotubularin related protein 2**
A. Bayram; K. Stumpfe; H. Wang; M. Pergande; H. Per; S. Çırak

P.168 **Falls in children and adolescents with Charcot-Marie-Tooth disease and typically developing children: a six-month prospective study**
K. Carroll; J. McGinley; K. Paterson; M. Ryan; R. Kennedy

P.169 **Aerobic anti-gravity exercise in patients with Charcot Marie Tooth disease: a pilot study**
K. Knak; L. Andersen; J. Vissing

P.170 **The effect of orthosis and foot surgery in a cohort of Charcot Marie Tooth disease children**
E. Milev; T. Bhandari; M. Laura; M. Reilly; F. Muntoni

P.171 ***CNTNAP1*: extending the phenotype of congenital hypomyelinating neuropathy in 6 further patients**
K. Low; K. Stals; R. Caswell; J. Clayton-Smith; A. Donaldson; N. Foulds; M. Splitt; A. Norman; K. Urankar; K. Vijayakumar; D. Study; S. Ellard; A. Majumdar; S. Smithson

P.172 **Homozygous p.R707W *MFN2* mutation is associated with neuropathy, lipomatosis, peripheral lipoatrophy and metabolic alterations**
M. Masingue; C. Vatier; I. Jéru; P. Latour; C. Jardel; P. Laforêt; B. Eymard; C. Vigouroux; T. Stojkovic

P.173 **Clinical characteristics of spectrum of *GNE* gene mutations in the Reunion-Island cohort**
I. Grigorashvili-Coin; M. Campech; F. Darcel; M. Jacquemont; M. Krahn; M. Cerino; A. Choumert

P.174 **Genotype-phenotype correlation analysis in *GNE* myopathy**
O. Pogoryelova; P. Cammish; H. Mansbach; H. Lochmüller

P.175 **Genetic characterization of a French cohort of *GNE*-mutation negative inclusion body myopathy patients using exome sequencing**
M. Cerino; S. Gorokhova; P. Laforêt; R. Ben Yaou; E. Salort-Campana; J. Pouget; S. Attarian; B. Eymard; J. Deleuze; A. Boland; A. Behin; T. Stojkovic; G. Bonne; N. Lévy; M. Bartoli; M. Krahn

P.176 ***GNE*-myopathy (HIBM): upper and lower extremity muscle strength declines over time in a prospective study**
A. Behin; T. Mozaffar; M. Tarnopolsky; T. Gidaro; O. Pogoryelova; J. Shah; S. Krolczyk; T. Koutsoukos; I. Tournev; H. Lochmüller

P.177 **Quantitation of sialylation status by lectin immunofluorescence in muscle biopsies of patients with *GNE* myopathy: assessing response to therapy**
M. Huizing; P. Leoyklang; B. Class; C. Ciccone; A. Glowacki; C. Jodarski; J. Perrault; W. Gahl; N. Carrillo; M. Malicdan

P.178 **A phase 3 randomized, double blind, placebo-controlled study to evaluate the efficacy and safety of sialic acid extended-release tablets in patients with *GNE* myopathy (GNEM)**
H. Lochmüller; A. Behin; Y. Caraco; H. Lau; M. Mirabella; I. Tournev; M. Tarnopolsky; O. Pogoryelova; J. Shah; T. Koutsoubos; A. Skrinar; H. Mansbach; E. Kakkis; T. Mozaffar

P.179 ***PLEC* gene mutations cause familial disto-proximal myopathy and long QT syndrome mimicking mitochondrial disease**
S. Servidei; G. Primiano; V. Muto; C. Cuccagna; D. Bernardo; D. Sauchelli; C. Sancricca; M. Lucchini; M. Mirabella; M. Tartaglia

P.180 **Adult onset distal myopathy secondary to nebulin gene mutations**
R. Juntas Morales; D. Lacourt; C. Theze; H. Pegeot; A. Perrin; K. Yauy; A. Maues de Paula; D. Figarella; N. Leboucq; M. Cossée

P.181 **Intramuscular injection of recombinant adeno-associated viral vectors expressing mutant *MATR3* recapitulates pathological features of VCPDM**
X. Zhang; S. Yamashita; N. Tawara; T. Doki; Z. Zhang; K. Hara; Y. Matsuo; M. Nagai; Y. Ando

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Chairpersons: Marianne de Visser and Werner Stenzel

P.182 **Immune-mediated necrotizing autoimmune myopathy: Dutch and Belgian experience**
M. de Visser; J. De Bleecker; A. van der Kooi; F. Eftimov; C. Saris; N. Voermans; J. Raaphorst; J. Lim; B. van Engelen

P.183 **Clinical and histopathological features of immune-mediated necrotising and inflammatory myopathy in relation to treatment with immune checkpoint blockers (ICBs) in cancer-patients**
S. Leonard Louis; M. Touat; T. Maisonobe; Y. Allenbach; O. Benveniste; D. Psimaras; T. Lenglet; O. Hadj Salem; N. Kramkimel; J. Cadranel; C. Lethrosne; J. Bruch; P. Laly; S. Knauβ; W. Stenzel

P.184 **The role of autophagy and protein homeostasis in immune-mediated necrotizing myopathy**
N. Fischer; C. Preuße; Y. Allenbach; O. Benveniste; H. Goebel; W. Stenzel

P.185 **First case report of nivolumab-induced dermatomyositis**
J. Bourgeois Vionnet; B. Joubert; E. Bernard; N. Fabien; M. Sia; V. Pante; J. Honnorat; N. Streichenberger

P.186 **Type 1 interferon signature as a diagnostic marker of dermatomyositis**
A. Uruha; Y. Allenbach; J. Charuel; L. Musset; A. Aussy; O. Boyer; K. Mariampillai; O. Landon-Cardinal; C. Rasmussen; S. Leonard-Louis; S. Suzuki; I. Nishino; W. Stenzel; O. Benveniste

P.187 **Dermatomyositis associated with hearing loss, peripheral nerve vasculitis and multi-system dysfunction**
P. Dhawan; E. Naddaf

P.188 **Anti-Mi2 dermatomyositis revisited: pure DM phenotype with muscle fiber necrosis and high risk of malignancy**
O. Landon-Cardinal; G. Monseau; Y. Schoindre; A. Rigolet; N. Champtiaux; B. Hervier; A. Masseau; E. Hachulla; T. Papo; B. Terrier; A. Meyer; F. Maurier; F. Gaches; E. Salort-Campana; S. Audia; A. Bouvier; W. Stenzel; O. Benveniste; B. Bienvenu; Y. Allenbach

P.189 **Incidence and prevalence of inclusion body myositis in western Sweden**
U. Lindgren; C. Lindberg; A. Oldfors

P.190 **Establishment of novel autoimmune animal model for sporadic inclusion body myositis**
N. Tawara; S. Yamashita; X. Zhang; Z. Zhang; T. Doki; Y. Matsuo; Y. Matsuo; S. Nakane; Y. Maeda; Y. Ando

P.191 **Mitochondrial analysis in wild-type TDP-43 transgenic mice mimicking sporadic inclusion body myositis**
Z. Zhang; S. Yamashita; N. Tawara; K. Kawakami; T. Doki; Y. Matsuo; X. Zhang; Y. Maeda; Y. Ando

P.192 **Muscle fiber dysfunction contributes to clinical muscle weakness in inclusion body myositis**
S. Lassche; A. Rietveld; A. Heerschap; J. Van Hees; M. Hopman; C. Saris; N. Voermans; B. Van Engelen; C. Ottenheijm

P.193 **Usefulness of cytoplasmic 5′-nucleotidase 1A autoantibodies for diagnosing patients with inclusion body myositis**
C. Ikenaga; M. Kadoya; A. Kubota; J. Shimizu

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S. Yamashita; Y. Matsuo; N. Tawara; X. Zhang; Z. Zhang; T. Doki; Y. Ando

P.195 **Impact of adipose-derived stem cells injection in a mouse model of inclusion body myositis**
V. Fabry; Q. Sastourné-Arrey; A. Girousse; Y. Jeanson; E. Uro-Coste; P. Cintas; L. Casteilla; C. Sengenès

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M. Johari; M. Arumilli; M. Savarese; J. Palmio; G. Tasca; M. Mirabella; L. Maggi; P. Hackman; B. Udd

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C. Gitiaux; C. Latroche; M. Weiss-Gayet; M. Rodero; D. Duffy; B. Bader Meunier; C. Bodemer; G. Mouchiroud; J. Chelly; S. Germain; I. Desguerre; B. Chazaud

P.198 **Clinical epidemiology and multidimensional analysis of idiopathic inflammatory myopathies: toward a classification based on myositis specific autoantibodies**
K. Mariampillai; B. Granger; M. Guiguet; D. Amelin; J. Charuel; L. Musset; Y. Allenbach; O. Benveniste

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W. Mauhin; K. Mariampillai; Y. Allenbach; L. Musset; J. Charuel; O. Benveniste

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S. Knauß; Y. Allenbach; C. Preuße; N. Fischer; V. Matyash; H. Goebel; O. Benveniste; W. Stenzel

P.201 **Differential type I and type II interferon signatures in primary inflammatory/dysimmune myopathies**
M. Rigolet; C. Hou; B. Periou; P. Muhammad; R. Gherardi; Y. Baba Amer; F. Authier

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J. Zamecnik; H. Storkanova; O. Krystufkova; M. Klein; H. Mann; L. Vernerova; M. Spiritovic; L. Senolt; J. Vencovsky; M. Tomcik

P.203 **Osmolyte accumulator expression is induced in muscle cells in response to inflammation**
B. De Paepe; J. Zschuntzsch; J. De Bleecker; J. Schmidt

P.204 **Osmolyte transporters of betaine GABA (SLC6A12) and taurine (SLC5A3) are expressed in muscle-infiltrating mononuclear cells in inflammatory myopathies**
B. De Paepe; J. Weis; J. De Bleecker

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T. Mozaffar; M. Wencel; N. Goyal; C. Philips; C. Olsen

P.206 **Anti-HMGCR antibody positive myopathy shows bcl-2-positive lymphocyte follicles**
T. Kurashige; N. Sumi; T. Kanbara; M. Ohta; T. Sugiura; H. Maruyama; T. Torii

P.207 **Macrophagic myofasciitis-associated cognitive dysfunction: a reappraisal of neuropsychological profile**
M. Aoun-Sebaiti; L. Danini; M. Derosin; P. Kauw; A. Bachoud-Levi; F. Authier

P.208 **Predictive value of cerebral FDG-PET for diagnosing aluminium hydroxide-induced macrophagic myofasciitis (MMF)**
A. Van der Gucht; M. Abulizi; M. Aoun-Sebati; E. Itti; F. Authier

P:209 **Inflammatory myopathy related to Chikungunya virus: A case report**
N. Muelas; P. Marti; I. Azorin; C. Gomis; J. Poyatos; M. Frasquet; J. Vazquez; L. Bataller; L. Gomez; R. Vilchez; T. Sevilla; J. Vilchez

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O. Ekmekci; G. Kavasoglu; M. Argin; A. Babaoglu; H. Karasoy; M. Korkmaz

P.211 **Cyclophosphamide, thalidomide, and dexamethasone as alternative treatment regimen for sporadic late onset nemaline myopathy associated with monoclonal gammopathy of undetermined significance**
J. Tanboon; T. Kumutpongpanich; W. Owattanapanich; T. Sangruchi; K. Boonyapisit; I. Nishino

Pompe disease (P.212–226)
Chairpersons: Tiziana Enrica Mongini and Benedikt Schoser

P.212 **Screening for late-onset Pompe disease among high-risk population in Japan**
K. Ogata; M. Kosuga; E. Takeshita; T. Matsumura; K. Ishigaki; S. Ozasa; H. Arahata; K. Sugie; T. Takahashi; S. Kuru; A. Hattori; H. Takada; M. Kobayashi; M. Takahashi; N. Tanaka; T. Okuyama; H. Komaki

P.213 **Identification of patients with late-onset Pompe disease (LOPD) based on muscle biopsy and clinical correlates: experience of a reference centre**
F. Lubieniecki; A. Taratuto

P.214 **Interim analysis of an investigator-initiated multi-site late-onset Pompe disease screening study**
M. Wencel; N. Goyal; T. Mozaffar

P.215 **Sensitivity and specificity of the PAS positive lymphocyte vacuoles in the diagnostic approach to late onset Pompe disease**
S. Sampaolo; S. Bernardini; A. Pascarella; O. Farina; C. Terracciano; L. Lombardi; F. Napolitano; T. Esposito; G. Di Iorio

P.216 **Functional and morphological improvement of skeletal muscle in Pompe disease after forced satellite cell activation**
G. Schaaf; T. van Gestel; S. In ‘t Groen; A. van der Ploeg; W. Pijnappel

P.217 **Stabilized next-generation recombinant human acid alpha-glucosidase ATB200 clears accumulated glycogen and reverses cellular dysfunction to increase muscle strength in a mouse model of Pompe disease**
S. Xu; Y. Lun; A. Nair; M. Frascella; A. Garcia; R. Soska; A. Ponery; A. Schilling; C. Della Valle; J. Feng; R. Gotschall; H. Do; K. Valenzano; R. Khanna

P.218 **Antisense oligonucleotides promote exon inclusion and correct the common c.-32-13T>G (IVS1) *GAA* splicing variant in iPS-derived skeletal muscle cells from Pompe patients**
W. Pijnappel; E. van der Wal; A. Bergsma; T. van Gestel; J. Pijnenburg; S. In ‘t Groen; H. Zaehres; M. Araúzo-Bravo; H. Schöler; A. van der Ploeg

P.219 **Long-term efficacy of enzyme-replacement therapy in 102 adult Pompe patients: a 5-year nationwide prospective cohort study**
L. Harlaar; E. Kuperus; M. Kruijshaar; S. Wens; J. de Vries; M. Favejee; J. van der Meijden; D. Rizopoulos; E. Brusse; P. van Doorn; A. van der Ploeg; N. van der Beek

P.220 **Antibody formation to enzyme replacement therapy in classic infantile Pompe disease: effects of immunomodulation in naïve patients**
E. Poelman; M. Hoogeveen-Westerveld; M. Kroos-de Haan; J. van den Hout; K. Bronsema; N. van de Merbel; A. van der Ploeg; W. Pijnappel

P.221 **Antibodies anti-ERT do not influence muscle fatty infiltration in a long cohort of patients with late onset Pompe disease**
S. Figueroa-Bonaparte; S. Segovia; I. Belmonte; I. Pedrosa; E. Montiel; J. Llauger; A. Alonso-Jimenez; E. Gallardo; I. Illa; J. Diaz-Manera

P.222 **Dilatative arterial malformations in patients with late onset Pompe disease (LOPD)**
A. Toscano; F. Granata; C. Rodolico; A. Ciranni; R. Arrigo; M. Longo; O. Musumeci

P.223 **Postural and gait patterns assessed by 3D movement analysis in a late onset Pompe disease sibship**
P. De Blasiis; D. Mazzoli; O. Farina; L. Lombardi; M. Melone; G. Di Iorio; S. Sampaolo

P.224 **Monitoring physical activity using a wearable device in Pompe disease**
A. Hamed; C. Curran; P. DasMahapatra

P.225 **Living with late-onset Pompe disease: the patient and clinician point of view**
N. Patel; S. Sathe; D. Dietze; C. Viereck; J. Barth; S. Sitaraman

P.226 **Late-onset limb-girdle myopathy with oculobulbar signs and rimmed vacuoles associated with a novel Pompe disease mutation**
M. Oliveira Santos; R. Taipa; M. Pires; I. Conceição

Periodic paralysis and related disorders (P.227–233)
Chairpersons: Nicol Voermans and Andoni J Urtizberea

P.227 **The Brody disease cohort study: clarification of the phenotype**
J. Molenaar; J. Verhoeven; N. Voermans; J. Mathieu; G. Vattemi; J. Franques; T. Kuntzer; L. Guyant-Marechal; S. Vicart; A. Behin; C. Erasmus; B. Brandom; E. Matthews; K. Suetterlin; B. van Engelen; D. Sternberg; B. Eymard

P.228 **Trifunctional protein beta subunit (*HADHB*) mutations associated with periodic paralysis phenotype**
G. Remiche; P. Baudin; C. Buon; J. Praline; K. Auré; M. Abramowicz; D. Sternberg; S. Nicole

P.229 **Skeletal muscle channelopathies: rare treatable disorders with common presentation in childhood**
E. Matthews; A. Silwal; R. Sud; A. Manzur; M. Hanna; F. Muntoni; P. Munot

P.230 **Large scale validation of functional expression of ClC-1 variants in genetic counselling of myotonia congenita**
K. Suetterlin; R. Sud; J. Burge; S. McCall; D. Fialho; A. Haworth; M. Sweeney; H. Houlden; S. Schorge; E. Matthews; M. Hanna; R. Mannikko

P.231 **Chronic progressive myopathy in a young patient with hyperkalemic periodic paralysis**
H. Shin; H. Jeong; H. Kim; J. Lee; Y. Choi

P.232 **Proximal myopathy without episodic weakness in relation with a probable novel mutation of the *CACNA1S* gene**
G. Moris; L. Martinez; A. Aurora; V. Alvarez

P.233 **Permanent muscle weakness in hypokalemic periodic paralysis**
S. Holm-Yildiz; N. Witting; F. Fornander; A. Eisum; M. Duno; T. Sorensen; J. Vissing

Biomarkers for dystrophinopathy (P.234–246)
Chairpersons: Sebahattin Cirak and Joel Schneider

P.234 **Expression pattern and biological function of miR-379 in muscular dystrophy**
M. Sanson; V. Mournetas; E. Massourides; I. Barthélémy; S. Blot; C. Pinset; I. Richard; D. Israeli

P.235 **Circulating miRs biomarkers for therapeutic monitoring in utrophin based DMD therapy**
N. Ramadan; S. Guiraud; B. Edwards; S. Squire; S. Hemming; K. Davies

P.236 **Establishment of a panel for the evaluation of the dystrophic process by quantitative RT-PCR**
C. Vaubourg; J. Bellec; E. Gicquel; W. Lostal; I. Richard

P.237 **MicroRNAs involved in nNOS regulation in dystrophic context**
M. Guilbaud; C. Gentil; I. Holtzmann; C. Gruszczynski; S. Falcone; C. Peccate; S. Benkhelifa-Ziyyat; S. Lorain; F. Aurade; L. Jeanson-Leh; F. Piétri-Rouxel

P.238 **Dystrophin Dp71 is expressed in skeletal muscle**
T. Kawaguchi; E. Niba; A. Rani; S. Yoshida; S. Sakakibara; N. Maeda; O. Sato; M. Matsuo

P.239 **Identification of serum protein biomarkers for utrophin based DMD therapy**
S. Guiraud; B. Edwards; S. Squire; A. Babbs; N. Shah; A. Berg; H. Chen; K. Davies

P.240 **Proteomic identification of novel brain and serum biomarkers linked to the pathophysiology of Duchenne muscular dystrophy**
S. Murphy; M. Zweyer; P. Dowling; M. Henry; P. Meleady; D. Swandulla; K. Ohlendieck

P.241 **Comparative high resolution proteomic analysis of dystrophic mouse models reveals a core dystrophic proteome and the impact of aging**
T. van Westering; H. Johansson; A. Coenen-Stass; S. Miyatake; J. Tanihata; S. Takeda; T. Yokota; J. Lehtiö; M. Wood; S. El Andaloussi; T. Roberts; Y. Aoki

P.242 **Longitudinal proteomic analysis of sera allows to non-invasively monitor disease progression in Duchenne muscular dystrophy**
P. Spitali; R. Tsonaka; K. Hettne; Z. Koeks; A. Roos; V. Straub; J. Piscos Domingos; F. Muntoni; C. Al-Khalili-Szigyarto; H. Lochmüller; E. Niks; A. Aartsma-Rus

P.243 **Urinary titin reveals persistent proteolysis in Duchenne muscular dystrophy**
H. Awano; M. Matsumoto; M. Nagai; T. Shirakawa; N. Maruyama; K. Iijima; Y. Nabeshima; M. Matsuo

P.244 **Urinary excretion of 8-OHdG, a biomarker of oxidative DNA damage, increases with age in DMD patients**
M. Matsumoto; H. Awano; M. Nagai; T. Shirakawa; K. Iijima; M. Matsuo

P.245 **Potential role of exosomes in skeletal muscle fibrosis**
S. Zanotti; S. Gibertini; F. Blasevich; S. Saredi; C. Bragato; A. Ruggieri; R. Mantegazza; L. Maggi; M. Mora

P.246 **Characterization of molecular pathophysiology in muscular dystrophy by next generation RNA sequencing using DMD and CMD mouse models**
N. Yanay; M. Elbaz; J. Konikov; S. Elgavish; M. Rabie; S. Mitterani-Rosenbaum; Y. Nevo

Centronuclear myopathies (P.247–264)
Chairpersons: Anders Oldfors and Marc Bitoun

P.247 **New myotubular myopathy classification**
C. Lilien; M. Annoussamy; T. Gidaro; E. Gargaun; V. Chê; U. Schara; A. D'Amico; A. Daron; J. Cuisset; M. Mayer; A. Hernandez; C. Vuillerot; S. Fontaine; C. deLattre; R. Bellance; V. Biancalana; A. Buj-Bello; J-Y. Hogrel; H. Landy; L. Servais

P.248 **Myotubular myopathy and excitation contraction coupling: from pathomechanism(s) to therapy**
J. Dowling; N. Sabha; N. Maani; J. Volpatti; H. Gonorazky; K. Rezai; L. Groom; R. Dirksen

P.249 **X-Linked myotubular myopathy (XLMTM): phenotypic variability**
C. Ortez; D. Natera; J. Colomer; D. Itzep; M. Alarcón; A. Frongia; C. Jou; A. Codina; C. Jiménez-Mallebrera; L. Martorell; V. Biancalana; L. González; P. Gallano; A. Nascimento

P.250 **Longitudinal data of patients with myotubular myopathy enrolled in a European prospective and longitudinal natural history study**
M. Annoussamy; C. Lilien; T. Gidaro; E. Gargaun; V. Chê; U. Schara; A. D'Amico; A. Daron; J. Cuisset; M. Mayer; A. Hernandez; C. Vuillerot; S. Fontaine; C. de Lattre; R. Bellance; V. Biancalana; A. Buj-Bello; J-Y. Hogrel; H. Landy; L. Servais

P.251 **Expression of the neuropathy-associated *MTMR2* gene rescues MTM1-associated myopathy**
M. Raess; B. Cowling; D. Bertazzi; C. Kretz; B. Rinaldi; P. Kessler; S. Friant; J. Laporte

P.252 **High-throughput transcriptome analysis provides new indicators of gene therapy efficacy in XLMTM dogs**
J. Dupont; J. Guo; J. Gray; A. Buj-Bello; M. Childers; D. Mack

P.253 **Clinical orphan patient pool methodology estimates current patient pool in centronuclear myopathy**
I. Vandersmissen; G. Vander Stichele; V. Biancalana; L. Thielemans

P.254 **Severe X-linked myotubular myopathy with unexpected inheritance from the grandfather and identification of necklace fibers in an asymptomatic male**
C. Hedberg-Oldfors; K. Visuttijai; A. Topa; M. Tulinius; A. Oldfors

P.255 **X-linked myotubular myopathy: a report of five cases**
J. Coelho; T. Proença Santos; T. Moreno

P.256 **A multicenter, retrospective medical record review of patients with X-linked myotubular myopathy (XLMTM): the RECENSUS study**
A. Beggs; B. Byrne; S. de Chastonay; T. Haselkorn; I. Hughes; E. James; N. Kuntz; J. Simon; L. Swanson; M. Yang; Z. Yu; S. Yum; S. Prasad

P.257 **A novel *MTM1* mutation in a patient with X-linked myotubular myopathy**
T. Rosa; J. Domingues; C. Iwabe-Marchese; A. Martinez; E. Mansur; M. França Jr; A. Nucci

P.258 ***BIN1* founder mutation in the Spanish gypsy population is the most frequent cause of adult onset centronuclear myopathies in the south of Spain**
M. Cabrera-Serrano; E. Rivas-Infante; F. Mavillard; B. Morar; D. Comas; A. Carvajal; R. Avila; N. Muelas; M. Olivé; J. Diaz; E. Verges; N. Romero; J. Laporte; J. Vilchez; N. Laing; L. Kalaydjieva; C. Paradas

P.259 **Myotonic discharges in a cohort of centronuclear myopathies.**
J. Domingues; T. Rosa; C. Iwabe-Marchese; C. Martins Jr; A. Martinez; L. Queiroz; B. Pfeilsticker; M. França Jr; A. Nucci

P.260 **Pseudo-dominant inheritance of a novel homozygous *HACD1* mutation associated with congenital myopathy: the first Caucasian family**
A. Toscano; V. Emmanuele; M. Savarese; O. Musumeci; A. Torella; E. Conca; M. Moggio; V. Nigro; C. Rodolico

P.261 **Antisense targeting of dynamin 2 by intramuscular delivery of vivo-morpholinos rescues the pathology in a murine model of myotubular myopathy**
N. Danièle; C. Bogni; L. Julien; A. Piet; A. Vignaud; A. Buj-Bello

P.262 **Antisense oligonucleotide-mediated *Dnm2* knockdown delays myotubular myopathy in mice after a single injection**
S. Buono; C. Kretz; C. Koch; A. Robé; S. Guo; B. Monia; J. Laporte; L. Thielemans; B. Cowling

P.263 **Vacuolar necklace muscle fibers – a new variant?**
J. Rinnenthal; C. Dittmayer; K. Irlbacher; K. Hahn; I. Wacker; W. Stenzel; H. Goebel

P.264 **Satellite cell alteration in *DNM2*-related centronuclear myopathy**
C. Almeida; M. Bitoun; M. Vainzof

18:00–19:30 **Symposium 1**

Thursday 5 October 2017

08:30–18:30 **Conference desk opens**

07:30–08:30 **NMD Editorial Board Meeting** – Hôtel France & Chateaubriand

09:00–10:30 **Extramuscular manifestations in NMD**
Invited lectures (NG.I.4–6) – Auditorium Chateaubriand
Chairpersons: Susan Iannaccone and Kevin Flanigan

NG.I.4 **AAV-mediated gene therapy in the central nervous system of dystrophin-Dp71 deficient mouse**
A. Rendon; O. Vacca; C. Vaillend

NG.I.5 **Respiratory insight into muscular dystrophy and relation to clinical trials**
B. Fauroux

NG.I.6 **The heart is a muscle too: the cardiomyopathy of Duchenne muscular dystrophy**
L. Cripe

10:30–11:00 **Morning refreshments, exhibition and posters –** Grand Large room and Jacques Cartier rotunda

11:00–11:30 **Extra-muscular manifestations in NMD**
Invited lectures (NG.I.7) – Auditorium Chateaubriand
Chairpersons: Mayana Zatz and Francesco Muntoni

NG.I.7 **Heart in laminopathies**
A. Muchir

11:30–13:00 **New genes and diseases**
Oral Presentations (NG.O.9–14) – Auditorium Chateaubriand
Chairpersons: Mayana Zatz and Francesco Muntoni

NG.O.9 **Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy**
V. Schartner; N. Romero; S. Donkervoort; S. Treves; P. Munot; T. Pierson; I. Dabaj; E. Malfatti; I. Zaharieva; F. Zorzato; B. Eymard; A. Taratuto; A. Boland; J. Deleuze; V. Biancalana; S. Quijano-Roy; F. Muntoni; C. Bönnemann; J. Laporte

NG.O.10 ***CASQ1* mutations impair calsequestrin polymerization and cause tubular aggregate myopathy**
J. Böhm; X. Lornage; S. Zanotti; P. Cudia; C. Schneider-Gold; E. Malfatti; M. Mora; J. Laporte

NG.O.11 **Recessive mutations in the novel gene *MST01* cause early onset neuromuscular condition**
A. Sarkozy; I. Zaharieva; A. Nasca; C. Scotton; R. Selvatici; M. Neri; O. Magnusson; A. Gal; D. Weaver; A. Armaroli; M. Pane; G. Hajnóczky; C. Sewry; R. Phadke; A. Donati; E. Mercuri; M. Zeviani; F. Muntoni; D. Ghezzi; A. Ferlini

NG.O.12 **A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 C-methyltransferase deficiency**
M. Malicdan; T. Vilboux; B. Ben-Zeev; J. Guo; A. Eliyahu; B. Pode-Shakked; A. Dori; S. Kakani; S. Chandrasekharappa; C. Ferreira; N. Shelestovich; D. Marek-Yagel; H. Pri-Chen; I. Blat; J. Niederhuber; C. Toro; J. Deeken; T. Yardeni; D. Wallace; W. Gahl; Y. Anikster

NG.O.13 **Recessive mutation in *EXOSC9* disrupts the exosome complex resulting in a novel form of cerebellar hypoplasia/atrophy with early motor neuronopathy**
S. Donkervoort; J. Müller; E. Knierim; D. Bharucha-Goebel; S. Dyack; D. Burns; Y. Hu; L. Baker; D. Ezzo; M. Scavina; A. Foley; M. Schülke; M. Schülke; C. Bönnemann

NG.O.14 **A common dominant-negative *COL6A1* pseudo-exon insertion is skippable using splice-modulating oligonucleotides**
V. Bolduc; A. Foley; S. Donkervoort; Y. Hu; B. Cummings; M. Lek; A. Sarathy; K. Sizov; H. Degefa; R. Wagener; G. Hennig; E. Hanssen; S. Lamande; F. Muntoni; S. Wilton; D. MacArthur; C. Bönnemann

13:00–14:00 **Lunch, exhibition and posters**

14:00–15:30 **Symposium 2**

15:30–17:00 **Poster session 3: parallel sessions (P.265–385)** – Poster area

Myotonic dystrophy (P.265–279)
Chairpersons: Duygu Selcen and David Hilton-Jones

P.265 **Results of a Japanese nationwide survey on congenital myotonic dystrophy**
M. Shichiji; K. Ishigaki; K. Ishiguro; T. Sato; T. Murakami; T. Matsumura; M. Osawa; S. Nagata

P.266 **A large multicenter study of pediatric myotonic dystrophy type 1 for evidence-based management**
E. Lagrue; C. Dogan; M. De Antonio; G. Bassez; D. Hamroun; R. Gherardi

P.267 **Clinical variability in myotonic dystrophy type 1: a five-categories disease classification fits clinical but not brain complexity**
S. Baldanzi; C. Simoncini; G. Ricci; C. Angelini; G. Siciliano

P.268 **Cardiac diastolic dysfunction correlates with the CTG trinucleotide repeat length in ambulatory myotonic dystrophy 1**
J. Park; D. Park; J. Sohn; J. Shin

P.269 **Association between mutation size and cardiac involvement in myotonic dystrophy type 1: an analysis of the DM1 heart registry**
K. Wahbi; C. Chong-Nguyen; V. Algalarrondo; H. Becane; P. Arnaud; D. Furling; G. Bassez; A. Behin; A. Fayssoil; P. Laforêt; T. Stojkovic; B. Eymard; D. Duboc

P.270 **Survival in myotonic dystrophy type 1 predicted by the new DM1 survival risk score**
K. Wahbi; R. Porcher; P. Laforêt; A. Fayssoil; T. Stojkovic; S. Leonard Louis; A. Behin; D. Furling; P. Arnaud; M. Sochala; V. Probst; D. Babuty; S. Pellieux; G. Bassez; Y. Pereon; B. Eymard; D. Duboc

P.271 **Venous thromboembolism in adult patients with inherited myopathies: a high risk in myotonic dystrophy**
K. Wahbi; M. Sochala; R. Porcher; T. Stojkovic; A. Behin; S. Leonard Louis; P. Laforêt; G. Bassez; B. Eymard; D. Duboc

P.272 **What is the prognosis after invasive ventilation for adult patients with a myotonic dystrophy type 1?**
C. Tard; E. Jaillette; A. Duva Pentiah; T. Perez; A. Mallart; S. NGuyen; A. Thevenon; V. Danel Brunaud; M. Preudhomme

P.273 **Early onset cataract: prominent feature in myotonic dystrophy type 2**
C. Papadopoulos; S. Xirou; K. Kekou; S. Kitsiou-Tzeli; E. Kararizou; G. Papadimas

P.274 **Post-receptor abnormalities contribute to peripheral insulin resistance in myotonic dystrophy type 1 and type 2**
L. Renna; F. Bosé; B. Fossati; M. Cavalli; G. Meola; R. Cardani

P.275 **Getting ready for clinical trials in myotonic dystrophy type 1 with the validation of functional outcome measures**
A. Jimenez-Moreno; N. Nikolenko; D. Moat; A. Mayhew; C. Massey; G. Gorman; J. Newman; M. Kierkegaard; A. Atalaia; C. Turner; H. Lochmüller

P.276 **Muscular fat fraction correlates with functionality in myotonic dystrophy type 1**
L. Heskamp; M. van Nimwegen; G. Bassez; A. Jimenez-Moreno; M. Ploegmakers; J. Deux; G. Gorman; H. Lochmüller; M. Catt; B. van Engelen; A. Heerschap

P.277 **Myd & Eat: a multidisciplinary intervention for myotonic dystrophy regarding food intake and nutrition**
S. van Hees; S. Knuijt; H. Dicke; T. Satink; J. Raaphorst; J. Groothuis; E. Cup

P.278 **CRISPR/Cas9-mediated genome editing corrects splicing alterations in myotonic dystrophy type 1**
M. Lo Scrudato; K. Poulard; A. Klein; S. Tomé; S. Martin; G. Gourdon; D. Furling; A. Buj-Bello

P.279 **Development of a MBNLΔ decoy-based gene therapy for myotonic dystrophy**
M. Matloka; L. Arandel; F. Rau; J. Marie; A. Ferry; N. Sergeant; D. Furling

Nemaline myopathy (P.280–296)
Chairpersons: Carina Wallgren Pettersson and Caroline Sewry

P.280 **The natural history of nemaline myopathy**
K. Amburgey; S. Hewson; C. Hum; M. Glueck; N. Sultanum; M. Girdea; M. Brudno; C. Bönnemann; J. Dastgir; R. Amin; O. Mayer; A. Constantinescu; F. Syed; T. Tran; M. Jain; L. Nelson; A. Beggs; C. Genetti; M. Lawlor; J. Dowling

P.281 **A novel copy number variation detection array for the diagnostics of neuromuscular disorders**
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P.377 **Olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a placebo-controlled phase 2 trial including a long-term, open-label follow-up study**
F. Muntoni; J. Buchbjerg; E. Bertini; E. Dessaud; E. Mercuri; J. Kirschner; C. Reid; A. Lusakowska; G. Comi; J. Cuisset; J. Abitbol; B. Scherrer; E. Vianna; W. van der Pol; C. Vuillerot; K. Gorni; P. Fontoura

P.378 **Efficacy and safety of nusinersen in children with later-onset spinal muscular atrophy (SMA): end of study results from the phase 3 CHERISH study**
E. Mercuri; R. Finkel; J. Kirschner; C. Chiriboga; N. Kuntz; P. Sun; S. Gheuens; C. Bennett; E. Schneider; W. Farwell; for the CHERISH Study Group

P.379 **Infants and children with SMA treated with nusinersen in clinical trials: an integrated safety analysis**
E. Mercuri; R. Finkel; M. Farrar; S. Richman; R. Foster; S. Hughes; W. Farwell; S. Gheuens

P.380 **Safety and efficacy of nusinersen in infants/children with spinal muscular atrophy (SMA): part 1 of the phase 2 EMBRACE study**
G. Acsadi; P. Shieh; T. Crawford; R. Richardson; N. Natarajan; D. Castro; S. Gheuens; G. Gambino; P. Sun; S. Reyna; W. Farwell; W. Mueller-Felber

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P.384 **Outcomes after 1-year in presymptomatic infants with genetically diagnosed spinal muscular atrophy (SMA) treated with nusinersen: interim results from the NURTURE study**
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F. Gattazzo; B. Laurent

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P.390 **Generation of biocompatible human artificial 3D skeletal muscle tissue from healthy and dystrophic pluripotent stem cells**
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P.392 **Analytical validation (based on CLIA & CLSI standards) of utrophin-laminin α2 and MHCd-laminin α2 duplex immunohistochemical assays using Computational Tissue Analysis (cTA™) for evaluation of Duchenne muscular dystrophy therapeutics**
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P.402 **WVE-210201, an investigational stereopure oligonucleotide therapy for Duchenne muscular dystrophy, induces Exon 51 skipping and dystrophin protein restoration**
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T. Meier; M. Leinonen; G. Buyse

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V. Selby; G. Ramdharry; J-Y. Hogrel; E. Milev; M. Hanna; F. Muntoni

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J. Pitchforth; M. Iodice; M. Main; S. Dziemian; A. Faisal; A. Bergsma; F. Muntoni

P.468 **Are muscle volume and echo-intensity related to rate of force development in children with Duchenne muscular dystrophy?**
M. Goudriaan; S. Schless; F. Cenni; M. van den Hauwe; N. Goemans; K. Desloovere

P.469 **Collection of high quality muscle biopsies for use in DMD clinical trial analysis; process development and implementation**
J. Tinsley; D. Frank; J. Dworzak; C. Faelan; J. Patterson-Kane; H. Wolff; F. Muntoni; PhaseOut DMD Study Group

***RYR1*-related myopathies and titinopathies** (P.470–484)
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J. Palmio; S. Leonard; S. Sacconi; M. Savarese; A. Semmler; J. Bach; W. Kress; T. Mozaffar; T. Lai; T. Stojkovic; B. Schoser; M. Walter; R. Reisin; A. Berardo; S. Attarian; A. Urtizberea; F. Fatehi; P. Hackman; B. Udd

P.471 **Adult onset recessive titinopathy with EDMD-like phenotype mimicking an acquired myositis**
S. Kapetanovic; L. Varona; K. Septien; M. Fernandez; E. Gallardo; M. Idoate; M. Garcia Barcina

P.472 **Unexpected gene expression findings in the titinopathy mouse model FINmaj-KI using RNA-Seq**
R. Krahe; I. Richard; B. Peng; P. Hackman; B. Udd; P. Jonson

P.473 **Do titin developmental isoforms contribute to the pathogenesis of congenital titinopathy?**
K. Yau; K. Jones; J. Smith; B. Cummings; M. Farrar; S. Cooper; M. Lek; E. Hoffman; V. Straub; A. Ferreiro; B. Udd; A. Beggs; C. Bönnemann; K. North; D. MacArthur; H. Granzier; F. Muntoni; M. Davis; N. Laing; E. Oates

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P. Jonson; S. Huovinen; P. Auvinen; P. Hackman; B. Udd; M. Savarese

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B. Lace; P. Gould; A. Dionne; N. Chrestian

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R. Vasavada; M. Waite; I. Chrismer; M. Jain; K. Meilleur; J. Witherspoon

P.479 **Morphological spectrum of *RYR1* recessive myopathies: clinical and genetic correlation.**
J. Rendu; E. Lacène; G. Brochier; M. Beuvin; C. Labasse; A. Madelaine; F. Levy Borsato; S. Vassilopoulos; J. Bevilacqua; F. Lubieniecki; S. Monges; A. Taratuto; M. Bitoun; G. Bonne; S. Sacconi; G. Antonini; N. Romero; M. Garibaldi

P.480 **Establishment of an international database of titin mutations and their phenotypes – a follow up**
M. Savarese; C. Bönneman; A. Ferreiro; A. Beggs; M. Gautel; M. Davis; T. Evangelista; J. Nikodinovic Glumac; J. Laporte; J. Smith; I. Richard; H. Granzier; R. Schneider; H. Jungbluth; S. Foye; A. Rockett Frase; B. Udd; P. Hackman

P.481 ***RYR1*-related myopathies: a wide range of clinical phenotypes and pathological histotypes**
A. Frongia; C. Ortez; D. Natera; D. Cortiza Itzep; M. Alarcón; M. Maioli; A. Maritza Betancourt Suarez; C. Jou; A. Codina; J. Corbera; L. Gonzalez; P. Gallano; J. Colomer; C. Jiménez-Mallebrera; A. Nascimento

P.482 **A novel approach to genotype-phenotype profiling for ryanodine receptor 1-related myopathies**
J. Witherspoon; C. Allen; S. Razaqyar; M. Cortes; I. Chrismer; K. Meilleur; M. Shelton

P.483 **Skeletal muscle oxidative stress is related to functional outcome measures in ryanodine receptor 1-related congenital myopathies**
J. Witherspoon; M. Razaqyar; I. Chrismer; A. Kuo; M. Shelton; C. Grunseich; A. Mankodi; A. Kokkinis; M. Waite; R. Vasavada; B. Drinkard; M. Jain; K. Meilleur; J. Todd

P.484 **Motor performance and disease progression in *RYR1*-RM**
J. Witherspoon; R. Vasavada; M. Waite; I. Chrismer; M. Jain; K. Meilleur

Managment and unusual observations (P.485–498)
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P.485 **European reference network for rare neuromuscular diseases: EURO-NMD**
R. Leary; M. Hails; H. Lochmüller; K. Bushby Newcastle University; T. Evangelista

P.486 **Audit of unplanned hospital admissions for patients with neuromuscular disorders in Cumbria and the north east of England**
E. Elliott; M. Guglieri; T. Evangelista; H. Lochmüller; V. Straub; K. Bushby; C. Marini-Bettolo

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D. Ho; V. Chê; A. Phelep; L. Servais; M. Annoussamy

P.488 **Using a traffic light system to aid advance care planning in neuromuscular patients**
C. Bassie; T. Willis; R. Kulshrestha; R. Vithlani

P.489 **Assessment of chewing function and investigation reliability of Karaduman chewing performance scale in pediatric neuromuscular diseases**
S. Serel Arslan; G. Aydın; I. Alemdaroğlu; O. Yılmaz; A. Karaduman

P.490 **Palliative medicine and neuromuscular disorders in adult's diseases and symptoms**
Y. Easthope-Mowatt; C. Bassie; M. McFarlene; R. Kulshresthra; T. Willis; D. Willis

P.491 **Walking with weakness: a snapshot of gait in a paediatric neuromuscular clinic**
K. de Valle; M. Ryan; A. Kornberg; R. Kennedy; K. Carroll

P.492 **Movement disorders hidden in the neuromuscular clinic**
S. Paus; J. Reimann

P.493 ***TOR1A* variants cause a severe arthrogryposis with developmental delay, strabismus and tremor**
A. Kariminejad; M. Dahl-Halvarsson; G. Ravenscroft; F. Afroozan; E. Keshavarz; H. Goullée; M. Davis; N. Laing; H. Tajsharghi

P.494 **A rare cause of congenital ptosis with external ophthalmoplegia: case report and differential diagnosis**
O. Kaiser; K. Rupprich; H. Kölbel; A. Della Marina; U. Schara; I. Sanchez-Albisua

P.495 **Epidemiological population-based study of hereditary myopathies in a southern European region**
I. Pagola; E. Vicente; L. Torne; E. Ardanaz; M. Ramos-Arroyo; I. Jerico; R. Fernandez-Torron

P.496 **Axial myopathy: clinical and histopathological features in 7 patients**
S. Choi; S. Ahn; S. Lim; J. Sung; J. Shin

P.497 **Axial myopathy in patients with neuromuscular diseases**
K. Knak; N. Witting; J. Vissing; K. Rudolf

P.498 **Dystrophin Dp427 is lost due to multiple DMD intron retentions in rhabdomyosarcoma CRL-2061 cells**
R. Yamanaka; M. Abdul Qawee; H. Awano; M. Matsumoto; H. Nishio; M. Matsuo; E. Niba

Friday 6 October 2017

07:45–14:15		**Conference desk opens**
08:15–09:45		**Advances in the treatment of neuromuscular disorders** Invited lectures (TH.I.8–10) – Auditorium Chateaubriand *Chairpersons: Melissa Spencer and Jerry Mendell*
	TH.I.8	**Antisense oligonucleotide therapies for neuromuscular disorders: where do we stand?** A. Aartsma-Rus
	TH.I.9	**Development of microdystrophins for gene therapy of DMD** J. Chamberlain; J. Ramos; K. Hollinger; J. Crudele; N. Bengtsson; S. Hauschka
	TH.I.10	**Neuromuscular disorders genetics: what is the best that we can do?** N. Laing
09:45–10:15		**Morning refreshments, exhibition and posters –** Grand Large room and Jacques Cartier rotunda
10:15–12:15		**New therapies and pathways** Oral presentations (TH.O.15–22) – Auditorium Chateaubriand *Chairpersons: Mariz Vainzof and Nicolas Lévy*
12:15–12:45		**Refreshment break**
	TH.O.15	**Gene therapy for oculopharyngeal muscular dystrophy** A. Malerba; P. Klein; H. Bachtarzi; S. Jarmin; G. Cordova; A. Ferry; V. Strings; M. Polay Espinoza; K. Mamchaoui; S. Blumen; J. Lacau St Guily; V. Mouly; M. Graham; G. Butler-Browne; D. Suhy; C. Trollet; G. Dickson
	TH.O.16	**A new AAV10-mediated gene therapy for *SOD1*-linked ALS** M. Biferi; M. Cohen-Tannoudji; A. Cappelletto; B. Giroux; M. Roda; S. Astord; T. Marais; A. Ferry; T. Voit; M. Barkats
	TH.O.17	**Whole-body rescue of Pompe disease with AAV liver delivery of engineered secretable *GAA* transgenes** P. Colella; F. Puzzo; M. Biferi; D. Bali; N. Paulk; P. Vidal; F. Collaud; M. Simon-Sola; S. Charles; R. Hardet; C. Leborgne; P. Sellier; L. van Wittenberghe; F. Boisgerault; M. Barkats; P. Laforêt; M. Kay; D. Koeberl; G. Ronzitti; F. Mingozzi
	TH.O.18	**Adeno associated vector-based gene therapy strategy for type 3 glycogen storage disease** P. Vidal; G. Ronzitti; F. Collaud; M. Simon Sola; P. Collela; F. Puzzo; H. Costa Verdera; S. Charles; A. Vignaud; L. Van Wittenberghe; B. Gjata; M. Gjorgjieva; P. Laforêt; F. Rajas; E. Malfatti; G. Comi; F. Mingozzi
	TH.O.19	**Tamoxifen increases survival, improves motor function and reduces levels of BIN1 and DNM2 in a mouse model of X-linked centronuclear (myotubular) myopathy** E. Gayi; H. Ismail; B. Cowling; L. Neff; J. Laporte; L. Scapozza; O. Dorchies
	TH.O.20	**Exhaustive characterization of the newly developed Duchenne muscular dystrophy rat model: a unique animal model for DMD which mimics the human disease at both the muscular and the cardiac levels** C. Huchet; G. Toumaniantz; T. Larcher; B. Fraysse; A. Lafoux; S. Remy; D. Caudal; M. Allais; E. Amosse; I. Anegon; C. Le Guiner
	TH.O.21	**Connexin-based hemichannels are key factors in the pathological mechanism underlying dysferlinopathy** G. Fernández; J. Bevilacqua; A. Cardenas; J. Sáez; P. Caviedes; L. Cea
	TH.O.22	**Prion-like protein aggregation of desmin myofibrillar myopathies** C. Weihl; J. Bieschke
12:45–14:15		**Symposium 3**
14:15–17:00		**Lunch bags and poster viewing**
19:30–00:30		**Congress dinner –** Quai Saint Malo

Saturday 7 October 2017

08:30–17:30 Conference desk opens

09:00–10:30 **New insights into muscle function, imaging, therapy and prevention**
Oral presentations (NI.O.23–27) – Auditorium Chateaubriand
Chairpersons: Gillian Butler-Browne and Haluk Topaloğlu

NI.O.23 **Sh3kbp1 involvement during skeletal muscle fibers formation: a new candidate for centronuclear myopathies**
A. Guiraud; N. Couturier; V. Buchman; A. Durieux; D. Arnould; E. Christin; S. Janczarski; M. Bitoun; V. Gache

NI.O.24 **Centronuclear myopathy-causing mutations in dynamin-2 impair actin-dependent trafficking in muscle cells**
A. González-Jamett; X. Baez-Matus; M. Bui; P. Guicheney; N. Romero; P. Caviedes; M. Bitoun; J. Bevilacqua; A. Cárdenas

NI.O.25 **Dynamic assessment of muscle perfusion, deoxymyoglobin and phosphorylated metabolites concentrations through fast interleaved NMR acquisitions with a clinical 3T scanner**
A. Lopez Kolkovsky; B. Marty; B. Coppa; E. Giacomini; P. Carlier

NI.O.26 **Brain imaging indicates genotype-phenotype association in Duchenne muscular dystrophy**
N. Doorenweerd; C. Bettolo; K. Hollingsworth; J. Hendriksen; E. Niks; V. Straub; H. Kan

NI.O.27 **MuSK MG patients showed a positive response to amifampridine phosphate in a randomized, placebo-controlled, crossover study**
R. Mantegazza; B. Pasanisi; C. Antozzi; L. Maggi; F. Andreetta; O. Simoncini; S. Bonanno

10:30–11:00 **Morning refreshments, exhibition and posters –** Grand Large room and Jacques Cartier rotunda

11:00–12:30 **Poster Highlights** – Auditorium Chateaubriand
Chairpersons: Beril Talim and Volker Straub

12:30–13:30 **WMS General Assembly –** Auditorium Chateaubriand

13:30–14:30 **Lunch, exhibition and posters**

14:30–16:30 **Late breaking session –** Auditorium Chateaubriand
Chairpersons: Alessandra Ferlini and John Vissing

16:30–17:30 **Prize giving and welcome to the 23rd WMS Congress**
Handover of the WMS flag and close of congress – Auditorium Chateaubriand