

## 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

- 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. Hourd ; L. F asson; 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. Ghimbovski; 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. Vrijssen; 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. Alemdarođlu; A. Karaduman; H. Topalođlu; 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. Alemdarođlu; H. Topalođlu; 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  $\alpha$ -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. Urtizbera; 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. Jhavar; 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 *LONPI*-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. Wehl
- 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. Ruparella; 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

- P87 **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
- P88 **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
- P89 **<sup>31</sup>P and <sup>1</sup>H nuclear magnetic resonance spectroscopy characterization of skeletal muscle pH dysregulation in Duchenne muscular dystrophy patients at rest**  
H. Reyngoudt; S. Turk; P. Carlier
- P90 **Nuclear magnetic resonance relaxometry characterization of D2-mdx mice**  
A. Martins-Bach; E. Araujo; B. Matot; Y. Fromes; P. Baudin; I. Richard; P. Carlier
- P91 **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
- P92 **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
- P93 **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
- P94 **Stimulated echo DTI of skeletal muscle in Becker muscular dystrophy: a pilot study**  
C. Baligand; J. Burakiewicz; M. Hooijmans; O. Scheidegger; M. Hall; P. Porcari; P. Carlier; C. Clark; A. Blamire; J. Verschuuren; E. Niks; H. Kan
- P95 **Skeletal muscle tissue characterization of a large cohort of patients with Becker muscular dystrophy using quantitative NMR imaging**  
B. Marty; M. Toussaint; R. Gilles; K. Wahbi; P. Carlier
- P96 **Effects of water compartmentation and distribution on skeletal muscle T1 values assessed by quantitative NMR imaging**  
B. Marty; B. Coppa; P. Baudin; P. Carlier
- P97 **Simple and fast drawing of regions of interest in leg muscles NMR images**  
P. Baudin; M. Beyeler; P. Carlier; O. Scheidegger
- P98 **Monitoring skeletal muscle chronic fatty degenerations using fast NMR T1-mapping**  
B. Marty; B. Coppa; P. Baudin; P. Carlier
- P99 **Reliability of DTI-based muscle-volumetry as compared to conventional T1-based manual segmentation**  
R. Rehmann; L. Schlaffke; R. Kley; M. Vorgerd; M. Tegenthoff
- P100 **Auto calculation of muscle impairment ratio utilizing Mercuri grades from CT and MR images of muscle**  
T. Nakayama; A. Ishiyama; T. Murakami; E. Kimura; S. Kuru
- Psycho-social aspects (P.101–117)**  
*Chairpersons: Isabelle Desguerre and Hanns Lochmüller*
- P101 **Audit of the TREAT-NMD global DMD and SMA registries: new insights into data collection methods**  
R. Leary; A. Oyewole; N. Goemans; H. Dawkins; C. Campbell
- P102 **Registry of congenital neuromuscular disorders in Japan: establishment and implementation**  
A. Ishiyama; E. Kimura; H. Nakamura; H. Komaki; M. Sasaki; I. Nishino



- P103 **Utilization of corticosteroids in DuchenneConnect registry participants**  
L. Cowen; M. Mancini; A. Lucas; A. Martin; J. Lavigne; J. Donovan
- P104 **Data analysis of dystrophinopathy national registry in Japan**  
E. Kimura; M. Mori-Yoshimura; H. Nakamura; H. Komaki; I. Nishino; S. Takeda
- P105 **DMD Genetic registry in Russia**  
D. Vlodayets; D. Reshetov; S. Artemieva; I. Shulyakova; O. Shidlovskaya; A. Monakhova; F. Vitrensky; D. Kazakov; E. Litvinova; E. Belousova
- P106 **An overview of the Cure SMA membership database: highlights of key demographic and clinical characteristics of registrants**  
L. Belter; T. Crawford; J. Jarecki; C. Jones; J. Kissel; M. Schroth; K. Hobby
- P107 **SMA registry in Russia**  
D. Vlodayets; D. Reshetov; O. Germanenko; S. Artemieva; I. Shulyakova; O. Shidlovskaya; A. Monakhova; F. Vitrensky; D. Kazakov; E. Litvinova; A. Tikhonov; E. Belousova
- P108 **Czech national registry of facioscapulohumeral muscular dystrophy**  
S. Vohanka; O. Parmova; L. Fajkusova; R. Mazanec; J. Strenkova; P. Ridzon; E. Ehler; M. Forgac; J. Junkerova; J. Haberlova; J. Stanek
- P109 **Myotubular/centronuclear myopathy patient registry: accelerating the pace of research and treatment**  
F. Takeuchi; J. Bullivant; K. Napier; M. Radochonski; L. Render; A. Hunter; M. Spring; A. Lennox; M. Bellgard; H. Lochmüller
- P110 **Long term follow-up of neuromuscular patients and family members submitted to psychoanalytical treatment**  
J. Forbes; T. Genesini; A. Mouzat; A. Bogochvol; D. Castro; D. Rüdiger; E. Padovan; E. Macedo; F. Fonseca; H. Andrade; H. Silva; L. Lise; L. Dantas; M. Naccache; T. Valladares; R. Pavanello; M. Zatz
- P111 **Patient's perception around disease progression and influential factors in spinal muscle atrophy**  
R. Muni Lofra; M. Eagle; A. Fort; D. Ramsey; M. Scotto; F. Muntoni; K. Bushby; H. Lochmüller; V. Straub; A. Mayhew
- P112 **Palliative medicine and neuromuscular disorders in adults – pilot symptom control clinic patient satisfaction and intervention efficacy**  
D. Willis; Y. Easthope-Mowatt; C. Bassie; M. McFarlane; R. Kulshresthra; T. Willis
- P113 **Referrals to psychology within a neuromuscular service: what do patients want to talk about?**  
J. Pattni; H. Clark; M. Desikan; R. Scalco; R. Quinlivan
- P114 **The influence of personal assistance on employment outcomes for young adults with neuromuscular disease**  
J. Wolff; L. Shaw
- P115 **Failure to attend neuromuscular clinic appointments: reasons and interventions**  
S. Cameron; G. Eglon; L. Hastings; V. Straub; H. Lochmüller; K. Bushby; M. Guglieri; C. Marini-Bettolo
- P116 **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
- P117 **Men with Duchenne muscular dystrophy and end of life planning**  
D. Abbott; H. Prescott; K. Forbes; J. Fraser; A. Majumdar

**Spinal muscular atrophy (P.118–133B)***Chairpersons: Ulrike Schara and Richard Finkel*

- 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
- P.119 **Abnormal fatty acid metabolism is a feature of spinal muscular atrophy**  
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
- P.121 **Nutritional status of a large cohort of children with spinal muscular atrophy type 2 (SMA2)**  
L. Schottlaender; M. Scoto; N. Imbrigiotta; T. Davis; M. Main; P. Munot; A. Sarkozy; A. Manzur; F. Muntoni
- 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. Viillard; 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 scapulooperoneal 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

- P133 **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
- P133B **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
- Laminopathies and dominant LGMD (P.134–149)**  
*Chairpersons: Andres Nascimento and Bjarne Udd*
- P134 **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
- P135 **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. Zanolati; K. Bushby; F. Muntoni; A. Rutkowski; C. Bönnemann; S. Quijano-Roy; G. Bonne; LMNA-rNHS Study Group
- P136 **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
- P137 **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. Kleinstaub; M. Lorenzo; A. Ishiyama; T. Sejersen; J. Gurgel-Giannetti; S. Monges; G. Bonne; S. Quijano-Roy
- P138 **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
- P139 **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
- P140 **Clinical and histopathological characterization of the first French case of *MATR3*-related distal myopathy**  
P. Laforêt; E. Malfatti; C. Metay; V. Jobic; R. Carlier
- P141 **Abnormal trafficking of connexin 43: a key element in the development of *LMNA* cardiomyopathy**  
C. Macquart; M. Chatzifrangkeskou; M. Gotthardt; G. Bonne; A. Muchir
- P142 **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
- P143 ***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
- P144 **Turnover studies on *DNAJB6* and the CASA pathway proteins**  
J. Sarparanta; S. Kawan; P. Jonson; B. Udd
- P145 **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
- Autosomal recessive LGMDs (P.150–166)**  
*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. Vandeveld; 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. Vandeveld; 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
- Charcot-Marie-Tooth disease and Distal myopathies (P.167–181)**  
*Chairpersons: Ichizo Nishino and Zohar Argov*
- 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 **CNTNAPI: 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
- Inflammatory myopathies (P.182–211)**  
*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 Kooij; 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. Knauß; W. Stenzel
- P.184 **The role of autophagy and protein homeostasis in immune-mediated necrotizing myopathy**  
N. Fischer; C. Preuß; 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. Masseur; 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



- P189 **Incidence and prevalence of inclusion body myositis in western Sweden**  
U. Lindgren; C. Lindberg; A. Oldfors
- P190 **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
- P191 **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
- P192 **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
- P193 **Usefulness of cytoplasmic 5'-nucleotidase 1A autoantibodies for diagnosing patients with inclusion body myositis**  
C. Ikenaga; M. Kadoya; A. Kubota; J. Shimizu
- P194 **CYLD is a possible therapeutic target for sporadic inclusion body myositis**  
S. Yamashita; Y. Matsuo; N. Tawara; X. Zhang; Z. Zhang; T. Doki; Y. Ando
- P195 **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
- P196 **Sporadic inclusion body myositis: a polygenic disorder?**  
M. Johari; M. Arumilli; M. Savarese; J. Palmio; G. Tasca; M. Mirabella; L. Maggi; P. Hackman; B. Udd
- P197 **Myogenic progenitor cells exhibit IFN type I-driven pro-angiogenic properties and molecular signature during juvenile dermatomyositis**  
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
- P198 **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
- P199 **Anti-mitochondrial antibodies are not a hallmark for severity in inflammatory myopathies**  
 W. Mauhin; K. Mariampillai; Y. Allenbach; L. Musset; J. Charuel; O. Benveniste
- P200 **PD1 and PDL2 axis confers T cell exhaustion in anti-SRP<sup>+</sup> and anti-HMGCR<sup>+</sup> myopathies**  
 S. Knauß; Y. Allenbach; C. Preuß; N. Fischer; V. Matyash; H. Goebel; O. Benveniste; W. Stenzel
- P201 **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
- P202 **Idiopathic inflammatory myopathies – increased expression of heat shock protein-90 in muscle tissue and plasma correlates with disease activity and skeletal muscle involvement**  
J. Zamecnik; H. Storkanova; O. Krystufkova; M. Klein; H. Mann; L. Vernerova; M. Spiritovic; L. Senolt; J. Vencovsky; M. Tomcik
- P203 **Osmolyte accumulator expression is induced in muscle cells in response to inflammation**  
B. De Paepe; J. Zschuntzsch; J. De Bleecker; J. Schmidt
- P204 **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
- P205 **Kv1.3 expression on effector memory T cells in sporadic inclusion body myositis: potential for targeted immunotherapy with dalazatide**  
T. Mozaffar; M. Wencel; N. Goyal; C. Philips; C. Olsen
- P206 **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
- P:210 **Parasitic myositis due to *Toxocara spp.* infection**  
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

- P221 **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
- P222 **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
- P223 **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
- P224 **Monitoring physical activity using a wearable device in Pompe disease**  
A. Hamed; C. Curran; P. DasMahapatra
- P225 **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
- P226 **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 (P227–233)**  
*Chairpersons: Nicol Voermans and Andoni J Urtizberea*
- P227 **The Brody disease cohort study: clarification of the phenotype**  
J. Molenaar; J. Verhoeven; N. Voermans; J. Mathieu; G. Vattermi; J. Franques; T. Kuntzer; L. Guyant-Marechal; S. Vicart; A. Behin; C. Erasmus; B. Bandom; E. Matthews; K. Suetterlin; B. van Engelen; D. Sternberg; B. Eymard
- P228 **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
- P229 **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
- P230 **Large scale validation of functional expression of CIC-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
- P231 **Chronic progressive myopathy in a young patient with hyperkalemic periodic paralysis**  
H. Shin; H. Jeong; H. Kim; J. Lee; Y. Choi
- P232 **Proximal myopathy without episodic weakness in relation with a probable novel mutation of the CACNAIS gene**  
G. Moris; L. Martinez; A. Aurora; V. Alvarez
- P233 **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*
- P234 **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
- P235 **Circulating miRs biomarkers for therapeutic monitoring in utrophin based DMD therapy**  
N. Ramadan; S. Guiraud; B. Edwards; S. Squire; S. Hemming; K. Davies
- P236 **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

- P237 **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
- P238 **Dystrophin Dp71 is expressed in skeletal muscle**  
T. Kawaguchi; E. Niba; A. Rani; S. Yoshida; S. Sakakibara; N. Maeda; O. Sato; M. Matsuo
- P239 **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
- P240 **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
- P241 **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
- P242 **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
- P243 **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
- P244 **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
- P245 **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
- P246 **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 (P247–264)**  
*Chairpersons: Anders Oldfors and Marc Bitoun*
- P247 **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
- P248 **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
- P249 **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
- P250 **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
- P251 **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

- P252 **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
- P253 **Clinical orphan patient pool methodology estimates current patient pool in centronuclear myopathy**  
I. Vandersmissen; G. Vander Stichele; V. Biancalana; L. Thielemans
- P254 **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
- P255 **X-linked myotubular myopathy: a report of five cases**  
J. Coelho; T. Proença Santos; T. Moreno
- P256 **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
- P257 **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
- P258 ***BINI* 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
- P259 **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
- P260 **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
- P261 **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
- P262 **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
- P263 **Vacuolar necklace muscle fibers – a new variant?**  
J. Rinnenthal; C. Dittmayer; K. Irlbacher; K. Hahn; I. Wacker; W. Stenzel; H. Goebel
- P264 **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 *MSTO1* 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**  
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 F. Ricci; R. Scalco; T. Mongini; G. Ferrero; A. Manole; E. Bertini; M. Desikan; I. Moroni; M. Di Rocco; H. Jungbluth; R. Quinlivan; H. Houlden
- P.370 **Exercise profile in patients with *SLC2A9* homozygous mutation and a history of exercise induced kidney failure**  
R. Scalco; A. Manole; S. Chatfield; S. Booth; R. Wigley; J. Pattni; Z. Michalak; M. Desikan; R. Godfrey; H. Houlden; E. Murphy; R. Quinlivan

**SMA therapy (P371–385)***Chairpersons: Enrico Bertini and Eugenio Mercuri*

- P371 **Safety and efficacy findings in the first-in-human trial (FIH) of the oral splice modulator branaplam in type 1 spinal muscular atrophy (SMA): interim results**  
L. Charnas; E. Voltz; C. Pfister; T. Peters; A. Hartmann; C. Berghs-Clairmont; J. Praestgaard; M. de Raspide; N. Deconinck; A. Born; G. Baranello; E. Bertini; U. Schara; N. Goemans; R. Roubenoff
- P372 **AVXS-101 phase 1 gene therapy clinical trial in SMA type 1: end-of-Study event free survival and achievement of developmental milestones**  
J. Mendell; S. Al-Zaidy; R. Shell; W. Arnold; L. Rodino-Klapac; T. Prior; L. Lowes; L. Alfano; K. Berry; K. Church; J. Kissel; S. Nagendran; J. L'Italien; D. Sproule; C. Wells; A. Burghes; K. Foust; B. Kaspar
- P373 **AVXS-101 phase 1 gene therapy clinical trial in SMA type 1: decreased need of ventilatory and nutritional support at End-of-Study**  
R. Shell; S. Al-Zaidy; W. Arnold; L. Rodino-Klapac; T. Prior; L. Lowes; L. Alfano; K. Berry; K. Church; J. Kissel; S. Nagendran; J. L'Italien; D. Sproule; C. Wells; A. Burghes; K. Foust; K. Meyer; S. Likhite; B. Kaspar; J. Mendell
- P374 **AVXS-101 phase 1 gene therapy clinical trial in SMA type 1: patients treated early with the proposed therapeutic dose were able to sit unassisted at a younger age**  
L. Lowes; S. Al-Zaidy; R. Shell; W. Arnold; L. Rodino-Klapac; T. Prior; L. Alfano; K. Berry; K. Church; J. Kissel; S. Nagendran; J. L'Italien; D. Sproule; C. Wells; A. Burghes; K. Foust; K. Meyer; S. Likhite; B. Kaspar; J. Mendell
- P375 **Clinical studies of RG7916 in patients with spinal muscular atrophy: SUNFISH part 1 study update**  
E. Mercuri; J. Kirschner; G. Baranello; L. Servais; N. Goemans; M. Pera; A. Marquet; T. Seabrook; S. Sturm; G. Armstrong; H. Kletzl; C. Czech; D. Kraus; H. Abdallah; L. Mueller; K. Gorni; O. Khwaja
- P376 **A series of case reports from JEWELFISH, an open-label study to investigate the safety, tolerability, and pharmacokinetics/pharmacodynamics of RG7916 in adult and pediatric patients with spinal muscular atrophy who previously participated in a study with another SMN2-targeting therapy**  
C. Chiriboga; E. Mercuri; D. Fischer; A. Marquet; D. Kraus; M. Alexander; A. Cho; G. Armstrong; H. Kletzl; C. Czech; T. Seabrook; H. Abdallah; L. Mueller; K. Gorni; O. Khwaja
- P377 **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
- P378 **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
- P379 **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
- P380 **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
- P381 **Nusinersen demonstrates greater efficacy in infants with shorter disease duration: end of study results from the ENDEAR study in infants with spinal muscular atrophy (SMA)**  
L. Servais; M. Farrar; R. Finkel; J. Kirschner; F. Muntoni; P. Sun; S. Gheuens; E. Schneider; W. Farwell
- P382 **Safety, tolerability and clinical efficacy of nusinersen in SMA type 1 older than 7 months: a prospective study**  
K. Aragon-Gawinska; E. Gargaun; A. Seferian; T. Gidaro; S. Gilabert; C. Lilien; C. Vuillerot; C. Cancès; A. Daron; E. Marucco; A. De Chalus; L. Flet Berliac; H. Armier; L. Fiedler; L. Servais

- P383 **First experience of Nusinersen early access program in patients with spinal muscular atrophy type 1**  
E. Gargaun; K. Aragon-Gawinska; A. Seferian; T. Gidaro; S. Gilabert; C. Lilien; A. Colcer; K. Boukouti; C. Vuillerot; C. Cancès; A. Daron; M. Annoussamy; A. De Chalus; L. Flet Berliac; H. Armier; L. Fiedler; L. Servais
- P384 **Outcomes after 1-year in presymptomatic infants with genetically diagnosed spinal muscular atrophy (SMA) treated with nusinersen: interim results from the NURTURE study**  
W. Hwu; D. De Vivo; E. Bertini; R. Foster; S. Gheuens; W. Farwell; S. Reyna
- P385 **Infants and children with SMA treated with nusinersen in clinical trials: experience of risk for respiratory or other events with repeat anesthesia/sedation for intrathecal administration**  
R. Finkel; E. Mercuri; C. Chiriboga; N. Kuntz; S. Richman; I. Bhan; S. Hughes; R. Foster; W. Farwell; S. Gheuens
- 16:15–16:45 **Afternoon refreshments, exhibition and posters – Grand Large room and Jacques Cartier rotunda**
- 17:00–18:30 **Poster session 4: parallel sessions (P. 386–498) – Poster area**
- Technical developments and stem cells (P386–395)**  
*Chairpersons: Miranda Grounds and Simone Spuler*
- P386 **Neuron-derived hiPSC: an *in vitro* model for the development of a gene therapy for myotonic dystrophy type 1**  
S. Ait Benichou; D. Jauvin; A. Arzoumanov; M. Varela; C. Bennett; M. Gait; M. Wood; J. Puymirat
- P387 **Patient-derived pluripotent stem cells: an *in vitro* model for neuromuscular diseases and high-throughput drug screening**  
S. Ait Benichou; L. Martineau; M. Chahine; P. Marquette; M. Maziade; A. Leblanc; J. Puymirat
- P388 **Engineering of a 3D bioartificial niche for adult satellite cells expansion: role of oxygen content**  
F. Gattazzo; B. Laurent
- P389 **Development of primary human satellite cells into an advanced therapeutic medicinal product (ATMP)**  
S. Spuler; A. Marg; J. Kieshauer; V. Schoewel; M. Vaegler
- P390 **Generation of biocompatible human artificial 3D skeletal muscle tissue from healthy and dystrophic pluripotent stem cells**  
S. Maffioletti; A. Henderson; S. Sarcar; I. Mannhardt; L. Moyle; M. Ragazzi; W. Wang; T. Eschenhagen; F. Tedesco
- P391 **Quantification of microdystrophin and correlation to circulating biomarkers**  
K. Brown; M. Lawlor; D. Golebiowski; P. Gonzalez; V. Ricotti; J. Schneider; C. Morris
- P392 **Analytical validation (based on CLIA & CLSI standards) of utrophin-laminin  $\alpha 2$  and MHCd-laminin  $\alpha 2$  duplex immunohistochemical assays using Computational Tissue Analysis (cTA™) for evaluation of Duchenne muscular dystrophy therapeutics**  
C. Faelan; J. Tinsley; A. Milici; S. Moore; J. Patterson-Kane
- P393 **Computational alignment of duplex immunohistochemically-stained muscle sections in support of therapies for Duchenne muscular dystrophy**  
L. Cerkovnik; J. Patterson-Kane; K. Ryall; A. Milici; J. Tinsley; S. Moore; C. Faelan
- P394 **Establishment of primary myoblast cell culture from cryoprotected skeletal muscle biopsies**  
B. Balci-Hayta; C. Bekircan-Kurt; E. Aksu; D. Dayangac-Erden; E. Tan; S. Erdem-Ozdamar
- P395 **Identification of new inhibitors of misfolded alpha-sarcoglycan degradation by high-throughput screening**  
L. Hoch; A. Egespipe; J. Marsolier; S. Henriques; I. Richard; X. Nissan

**Clinical drug development for DMD (P.396–413)***Chairpersons: Kay Davies and Mar Tulinius*

- P.396 **Edasalonexent (CAT-1004), an NF- $\kappa$ B inhibitor, enhances myotube formation *in vitro*, and increases exon-skipped sarcolemmal dystrophin in muscle of mdx mice**  
A. Nichols; J. Reilly; F. Liu; P. Bista; D. Lee; S. Webb; D. Picarella; J. Wood; M. Yao; M. Passini; N. Estrella
- P.397 **MoveDMD: phase 2 trial of edasalonexent, an NF- $\kappa$ B inhibitor, in 4 to 7-year old patients with Duchenne muscular dystrophy**  
R. Finkel; K. Vandeborne; H. Sweeney; E. Finanger; G. Tennekoon; P. Shieh; R. Willcocks; S. Forbes; W. Triplett; S. Yum; M. Mancini; M. Friedman; A. Fretzen; J. Donovan
- P.398 **Rimeporide: safety, tolerability and pharmacokinetic results from a phase Ib study in DMD boys as well as exploratory biomarkers**  
T. Gidaro; L. Servais; S. Previtali; A. Zamboni; J. Pitchforth; K. Maresh; J. Diaz; C. Laveille; J. Gray; F. Porte-Thomé; H. Gheit; M. Annoussamy; V. Che; D. Duchene; M. Sora; S. Gerevini; N. Vidal; K. Groves; J. Brimble; F. Muntoni
- P.399 **Effects of long-term eteplirsen treatment on upper limb function in patients with Duchenne muscular dystrophy: findings of two phase 2 clinical trials**  
L. Alfano; K. Berry; J. Mendell; H. Eliopoulos; L. Han; L. Lowes
- P.400 **Stunning pharmacological properties of DS-5141b, an antisense oligonucleotide consisting of 2'-O,4'-C-ethylene-bridged nucleic acids and 2'-O-methyl RNA, on dystrophin mRNA exon skipping**  
K. Takaishi; M. Kakuta; K. Ito; A. Kanda; H. Takakusa; H. Miida; T. Masuda; A. Nakamura; Y. Onishi; T. Onoda; Y. Kazuki; M. Oshimura; Y. Takeshima; M. Matsuo; M. Koizumi
- P.401 **Injection site reactions as a consequence of long-term subcutaneous administration of drisapersen in Duchenne muscular dystrophy**  
E. Niks; R. Van Doorn; J. Domingos; M. Guglieri; M. Pane; A. Mauro; A. Virgili; M. Morren; A. Martinez; A. Nguyen; M. Hooijmans; I. De Groot; A. Ferlini; M. Tulinius; V. Straub; F. Muntoni; J. Kirschner; E. Mercuri; N. Goemans
- P.402 **WVE-210201, an investigational stereopure oligonucleotide therapy for Duchenne muscular dystrophy, induces Exon 51 skipping and dystrophin protein restoration**  
M. Wood; J. Zhang; K. Bowman; D. Butler; C. Rinaldi; G. McClorey; M. Frank-Kamenetsky; N. Iwamoto; N. Kothari; G. Lu; S. Mathieu; M. Meena; S. Menon; M. Shimizu; S. Standley; H. Yang; Z. Zhong; C. Francis; C. Vargeese
- P.403 **PhaseOut DMD: a Phase 2, proof of concept, clinical study of utrophin modulation with ezutromid**  
F. Muntoni; K. Maresh; K. Davies; S. Harriman; G. Layton; R. Roskamp; A. Russell; B. Tejura; J. Tinsley
- P.404 **Drug development of vamorolone for Duchenne muscular dystrophy**  
P. Clemens; M. Guglieri; L. Morgenroth; J. Damsker; A. Smith; Y. Hathout; A. Cnaan; E. Smith; J. Mah; B. Byrne; D. Castro; R. Finkel; N. Kuntz; C. McDonald; Y. Nevo; M. Ryan; M. Tulinius; R. Webster; E. Hoffman; CINRG Investigators
- P.405 **A Phase I, single- and repeated-dose study of TAS-205, a novel inhibitor of hematopoietic prostaglandin D synthase, in patients with Duchenne muscular dystrophy**  
H. Komaki; E. Takeshita; Y. Motohashi; A. Ishiyama; M. Sasaki; K. Miyoshi; I. Yamamiya; N. Yamada; N. Minami
- P.406 **A phase 2 trial of the safety and pharmacokinetics of ataluren in patients aged 2 to 5 years with nonsense mutation Duchenne muscular dystrophy**  
P. Riebling; R. Kong; E. O'Mara; X. Luo; P. Trifillis; T. Ong
- P.407 **Design of a phase 3 trial to evaluate the long-term efficacy and safety of ataluren in patients with nonsense mutation Duchenne muscular dystrophy**  
P. Riebling; E. O'Mara; X. Luo; P. Trifillis; T. Ong

- P408 **Consistency of efficacy of idebenone in respiratory decline in Duchenne muscular dystrophy (DMD): comparison of analysis methods**  
T. Meier; M. Leinonen; G. Buyse
- P409 **Meta-analysis of two clinical trials with idebenone in patients with Duchenne muscular dystrophy (DMD): impact on respiratory decline**  
T. Meier; M. Leinonen; G. Buyse
- P410 **Impact of idebenone on pulmonary morbidity, including bronchopulmonary adverse events, in Duchenne muscular dystrophy**  
O. Mayer; C. Rummey; G. Buyse
- P411 **Repurposing tamoxifen for severe myopathies: from preclinical evaluation in animal models to clinical trials in patients**  
O. Dorchies; E. Gayi; H. Ismail; L. Neff; B. Cowling; J. Laporte; T. Dor; D. Fischer; U. Rüegg; L. Scapozza
- P412 **Daily versus weekend steroid use in DMD: age at loss of ambulation is equivalent in a retrospective patient cohort**  
M. Waldrop; J. Kaminoh; M. Moore-Clingenpeel; K. Flanigan; o. on behalf
- P413 **Cataract development associated with long term glucocorticoid therapy in DMD patients**  
M. Rice; M. Yang; P. Horn; J. Bange; B. Wong
- Neuromuscular transmission defects (P414–425)**  
*Chairpersons: Jiri Vajsar and Yoram Nevo*
- P414 **Recessively-acting choline transporter mutations associated with severe congenital myasthenia disrupt transporter surface trafficking *in vitro* and *in vivo***  
H. Wang; S. Salter; O. Refai; H. Hardy; T. Sejersen; J. Wright; H. Zimmerman; J. Weis; U. Schara; M. Russell; O. Abdul-Rahman; J. Chilton; R. Blakely; E. Baple; A. Crosby; S. Cirak
- P415 **Congenital myasthenia syndromes: clinical description of a pediatric cohort**  
A. Bénézit; D. Sternberg; S. Nicole; S. Bauché; C. Gitiaux; C. Barnerias; R. Rubinsztajn; J. Bergounioux; B. Mbieleu; C. Ioos; R. Sauvagnac; Y. Ivanovic; V. Coudert; H. Amthor; I. Dabaj; I. Dabaj; B. Estournet; S. Quijano-Roy
- P416 **A common *CHRNE* mutation (c.130dupG) in Brazilian patients with congenital myasthenic syndrome**  
E. Estephan; A. Silva; R. Mendonça; V. Caldas; A. Zambon; P. Marchiori; C. Heise; U. Reed; E. Zanoteli
- P417 **Genetic landscape of congenital myasthenic syndroms from Turkey: novel mutations and clinical insights**  
U. Yiş; K. Becker; S. Kurul; G. Uyanik; E. Bayram; G. Haliloglu; I. Polat; M. Ayanoglu; D. Okur; A. Tosun; G. Serdaroglu; S. Yılmaz; H. Topaloğlu; B. Anlar; S. Cirak; A. Engel
- P418 ***GFPT1*-related limb-girdle myasthenia: first case reported in Argentina**  
M. Rugiero; H. Gonorazky; M. Bettini; M. Saccoliti; X. Lornage; J. Böhm; J. Laporte; N. Romero; A. Taratuto
- P419 **Cataracts, cognitive impairment, and congenital myasthenic syndrome with myopathic features caused by mutation in *GMPPB* gene**  
A. Nascimento; C. Ortez; D. Natera; A. Frongia; M. Alarcon; D. Itzep; C. Jou; A. Codina; J. Corbera; C. Jimenez-Mallebrera; M. Rodriguez; L. González; P. Gallano; J. Colomer
- P420 **A novel mutation in *AGRN* gene causing congenital myasthenic syndrome with distal myopathy**  
A. Bamaga; M. Al-Lozi; C. Weihl
- P421 **A case of mistaken diagnosis with serendipitous therapeutic implications**  
N. Mc Sweeney; M. Carter; A. Greene; O. O Mahony; B. Lynch
- P422 **Development of a specific home based assessment tool for monitoring fluctuations in physical function and muscle performance in adult and paediatric myasthenia patients**  
V. Selby; G. Ramdharry; M. Hanna; F. Muntoni

- P423 **Functional fatigue in a sample of the UK myasthenic population**  
V. Selby; G. Ramdharry; J-Y. Hogrel; E. Milev; M. Hanna; F. Muntoni
- P424 **Tacrolimus treatment in patients with long-standing ocular myasthenia gravis**  
S. Oh
- P425 **Establishing a relationship between EQ-5D and QMG in patients with Lambert-Eaton myasthenic syndrome**  
J. Jarrett; R. Mantegazza; J. Sieb; J. Datt
- Outcome measures for neuromuscular diseases (P426–439)**  
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- P426 **New outcome measures for pain and fatigue during a typical day: pain and fatigue graphs**  
R. Janssen; E. Cup; T. Packer; J. Ijspeert; N. van Alfen; J. Groothuis
- P427 **Family impact and Health-Related Quality of Life (HRQoL) of parents and individuals with SMA**  
L. Belter; J. Jarecki; K. Hobby; M. Teynor
- P428 **Associations between NMR, electrophysiological, strength and function variables in SMA type 2 and 3**  
J-Y. Hogrel; M. Annoussamy; A. Chabanon; A. Daron; Y. Péréon; C. Cancès; C. Vuillerot; N. Goemans; J. Cuisset; V. Laugel; U. Schara; E. Gargaun; T. Gidaro; A. Seferian; S. Turk; R. Hermosilla; E. Fournier; P. Baudin; P. Carlier; L. Servais; NatHis SMA Study Group
- P429 **Innovative home activity monitoring in non-ambulant patients with spinal muscular atrophy: a multicenter observational trial**  
E. Gargaun; A. Seferian; G. Quicke; A. Moraux; T. Gidaro; E. Gasnier; A. Daron; Y. Péréon; C. Cancès; C. Vuillerot; J. Cuisset; E. Toledano; R. Hermosilla; O. Khwaja; C. Czech; A. Chabanon; M. Annoussamy; D. Vissiere; L. Servais
- P430 **Use of the Microsoft Kinect during motor function assessments of patients with Spinal muscular atrophy children: Kinect-MFM study**  
D. Vincent-Genod; J. Coton; P. Rippert; G. Thomann; C. Vuillerot
- P431 **Feasibility and validation of modified oculobulbar facial respiratory score (mOBFRS) in amyotrophic lateral sclerosis (ALS) and sporadic inclusion body myositis (sIBM)**  
M. Wencel; N. Araujo; T. Mozaffar; N. Goyal
- P432 **The gross motor function measure is valid for Fukuyama congenital muscular dystrophy**  
T. Sato; M. Adachi; K. Nakamura; M. Zushi; K. Goto; T. Murakami; K. Ishiguro; M. Shichiji; K. Saito; T. Ikai; M. Osawa; I. Kondo; S. Nagata; K. Ishigaki
- P433 **Evaluation of skeletal muscle in patients with Fukuyama congenital muscular dystrophy (FCMD) using bioelectrical impedance analysis**  
T. Murakami; K. Ishigai; K. Ishiguro; T. Sato; M. Shichiji; M. Ikeda; S. Nagata; T. Uchida; S. Kuru; T. Nakayama
- P434 **ActiMyo home monitoring in adult patients with limb girdle muscular dystrophy type 2B and facioscapulohumeral muscular dystrophy in study ATYR 1940-C-004**  
T. Gidaro; A. Moraux; M. Grelet; E. Gasnier; M. Villeret; M. Annoussamy; J. Vissing; S. Attarian; T. Mozaffar; S. Iyadurai; K. Wagner; G. Walker; A. Richiardi; S. Shukla; D. Vissière; L. Servais
- P435 **Validity of the 6-minute walking test in myotonic dystrophy type 1 in a large scale cross-sectional study**  
D. Moat; C. Jimenez-Moreno; A. Mayhew; C. Massey; N. Nikolenko; C. Turner; H. Lochmüller
- P436 **Associations between grip strength, myotonia and CTG expansion in myotonic dystrophy type 1**  
J-Y. Hogrel; G. Ollivier; I. Ledoux; L. Hébert; B. Eymard; J. Puymirat; G. Bassez



- P437 **Clinical outcome study of dysferlinopathy: what are the best outcome measures for dysferlinopathy patients?**  
M. James; M. Jacobs; A. Mayhew; J. Feng; 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
- P438 **Is grip strength compared to lower extremity measurements sufficient for capturing changes in muscle strength in chronic inflammatory demyelinating polyneuropathy?**  
K. Knak; L. Andersen; L. Markvardsen
- P439 **Optimization and implementation of best practices for collection and preparation of muscle biopsies for analysis during clinical trials of neuromuscular disease therapeutics**  
D. Frank; J. Dworzak; M. Lawlor; S. Lewis; Z. Sahenk; M. Stewart; C. Kincaid; C. Sewry; L. Feng; R. Phadke; F. Muntoni; J. Mendell; S. Moore
- Excitation–contraction coupling (P440–451)**  
*Chairpersons: Carmen Navarro and Ikuya Nonaka*
- P440 **Functional characterisation of p.Trp284Ser *STAC3* mutation causing impaired excitation-contraction coupling in congenital myopathy patients**  
I. Zaharieva; A. Sarkozy; A. Manzur; P. Munot; H. Jungbluth; L. Feng; R. Phadke; C. Sewry; S. Treves; F. Muntoni
- P441 **Tubular aggregate myopathy with dystrophic features**  
J. Lee; M. Yoshimura; R. Hirano; S. Miyatake; E. Koshimizu; N. Matsumoto; H. Mori; N. Tachii; M. Suzuki; K. Ogata; N. Ichizo; S. Noguchi
- P442 **Clinics, histopathology and whole-body-MRI pattern in *CACNA1S/DHPR* myopathy**  
Y. Ivanovic-Barbeito; I. Dabaj; E. Malfatti; A. Bénézit; H. Gonorazky; A. Taratuto; J. Laporte; B. Eymard; N. Romero; N. Pakleza; R. Carlier; S. Quijano-Roy
- P443 **Identification and characterisation of *ATP2A1* variants through whole exome sequencing**  
K. Johnson; A. Martinez Arroyo; M. Zulaica; R. Fernández-Torrón; A. Lopez de Munain; A. Töpf; M. Bertoli; L. Phillips; A. Blain; M. Ensini; M. Lek; T. Mullen; E. Valkanas; L. Xu; D. MacArthur; V. Straub
- P444 **Congenital myopathy associated with the *Triadin* knockout syndrome**  
A. Engel; K. Redhage; D. Tester; M. Ackerman; D. Selcen
- P445 **Calreticulin mutation in a case of myopathy**  
S. Sampaolo; D. de Lucia; I. Lombardi; S. Casertano; F. Rossi; M. Fratta; R. Di Francia; G. Di Iorio
- P446 **Slow relaxation kinetics of sarcomeres contribute to muscle slowness in *NEM6* patients**  
J. de Winter; J. Molenaar; S. Lassche; E. Malfatti; N. Romero; M. Persson; D. Rassier; B. van Engelen; N. Voermans; C. Ottenheijm
- P447 **From voltage sensing to gene expression in the control of muscle mass homeostasis**  
S. Falcone; C. Gentil; C. Benedetto; M. Traoré; A. Ferry; F. Piétri-Rouxel
- P448 **SPEG deficiency is associated with muscle weakness, triad defect, abnormal calcium handling and EC coupling**  
V. Huntoon; J. Widrick; C. Sanchez; C. Kutchukian; S. Cao; A. Beggs; V. Jacquemond; P. Agrawal
- P449 **Muscle growth by activin type II receptor blocking ameliorates weakness in *GNE* myopathy mice**  
M. Miyakawa; T. Yonekawa; M. Malicdan; E. Lach-Trifilieff; I. Nonaka; I. Nishino; S. Noguchi
- P450 **Myostatin expression levels in neuromuscular diseases participates in anti-myostatin clinical failure**  
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

- P451 **CXCL12 and osteopontin from bone marrow-derived mesenchymal stromal cells improve muscle regeneration by influencing upon muscle satellite cell and myoblast**  
Y. Maeda; Y. Yonemochi; Y. Nakajyo; H. Hidaka; T. Ikeda; Y. Ando  
**Outcome measures for DMD (P.452–469)**  
*Chairpersons: Laurent Servais and Jan Verschuuren*
- P452 **Accelerating the translation of natural history into more effective clinical trial design through multi-stakeholder collaboration**  
E. Mercuri; N. Goemans; F. Muntoni; A. Manzur; B. Wong; J. Signorovitch; S. Ward
- P453 **Consistency between natural history and clinical trial placebo arms for 48-week changes in six-minute walk distance (6MWD) in patients with Duchenne muscular dystrophy (DMD)**  
E. Mercuri; N. Goemans; G. Sajeev; Z. Yao; E. McDonnell; S. Ward; J. Signorovitch
- P454 **Validation of a prognostic score for changes in six-minute walk distance (6MWD) in patients with Duchenne muscular dystrophy (DMD)**  
N. Goemans; M. vanden Hauwe; G. Sajeev; Z. Yao; E. McDonnell; S. Ward; J. Signorovitch
- P455 **Outcome measures for Duchenne muscular dystrophy from ambulant to non-ambulant patients: Implications for clinical trials**  
J. Domingos; M. Eagle; A. Moraux; J. Butler; V. Decostre; D. Ridout; A. Mayhew; V. Selby; M. Guglieri; M. Van der Holst; M. Jansen; J. Verschuuren; I. de Groot; E. Niks; L. Servais; J-Y. Hogrel; V. Straub; T. Voit; V. Ricotti; F. Muntoni
- P456 **Development of novel observer-reported outcome assessments in clinical trials of patients with Duchenne muscular dystrophy**  
D. Martin; C. Macary; C. Jones; L. Walker; M. O'Connor; E. Thompson; K. Gallitano; M. Leffler; C. McSherry; M. Kosinski; M. White
- P457 **Development and sensibility evaluation of the muscular dystrophy child health index of life with disabilities questionnaire**  
R. Propp; S. Weir; C. Encisa; A. Davis; L. McAdam; N. Salbach; U. Narayanan
- P458 **Activities of daily living detection using home activity monitoring device in Duchenne muscular dystrophy patients**  
A. Moraux; B. Beaufiles; M. Grelet; A. Seferian; E. Gasnier; T. Gidaro; E. Dorveaux; D. Vissiere; L. Servais
- P459 **Hand function in boys and men with Duchenne muscular dystrophy (DMD)**  
M. Hunnekens; J. Huijben; I. de Groot
- P460 **Unraveling upper extremity performance in DMD: a biophysical model**  
M. Janssen; J. Harlaar; H. Koopman; I. de Groot
- P461 **Performance of the upper limb module for Chinese patients with Duchenne muscular dystrophy: a new useful clinical tool to monitor the disease progress and as an outcome measure for therapeutic drug trial**  
Y. Chiu; W. Choi; C. Hui; S. Li; S. Lee; S. Chan
- P462 **Exploring physical activity levels and sleep efficiency relationships among boys with Duchenne muscular dystrophy (DMD)**  
R. Bendixen; J. Anning; A. Kelleher; M. Yuhas; M. Feltman; D. Lorenzin; H. Morizono; E. Hoffman
- P463 **Longitudinal home-monitoring data in non-ambulant patients with Duchenne muscular dystrophy**  
A. Seferian; G. Quicke; E. Gargaun; A. Moraux; S. Gillabert; C. Lilien; E. Gasnier; V. Che; T. Gidaro; M. Annoussamy; D. Vissiere; L. Servais
- P464 **The 100-meter timed test: ability to detect change over time in Duchenne muscular dystrophy**  
N. Miller; L. Alfano; K. Flanigan; S. Al-Zaidy; C. Tsao; J. Mendell; L. Lowes
- P465 **The reliability and validity of Turkish version of pedsQL multidimensional fatigue scale in Duchenne muscular dystrophy**  
I. Alemdaroğlu; N. Bulut; S. Bozgeyik; A. Karaduman; H. Topaloğlu; O. Yılmaz

- P466 **Finding clinical meaning in patient-reported functional health: development of the Duchenne muscular dystrophy lifetime mobility scale**  
E. Henricson; C. McDonald; A. Mayhew; A. Bagley; N. Joyce; B. Oskarsson; L. Sodeberg-Miller; S. Liu; R. Abresch; CINRG Investigators
- P467 **A clinical update on the eNHANCE project: eye tracking control for reaching and grasping in an adolescent Duchenne muscular dystrophy (DMD) population**  
J. Pitchforth; M. Iodice; M. Main; S. Dziemian; A. Faisal; A. Bergsma; F. Muntoni
- P468 **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
- P469 **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 (P470–484)**  
*Chairpersons: Peter Hackman and John Rendu*
- P470 **Expanding importance of HMERF titinopathy: new mutations and clinical aspects**  
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. Urtizbera; F. Fatehi; P. Hackman; B. Udd
- P471 **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
- P472 **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
- P473 **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. Ferreira; B. Udd; A. Beggs; C. Bönnemann; K. North; D. MacArthur; H. Granzier; F. Muntoni; M. Davis; N. Laing; E. Oates
- P474 **Rare phenotypes related to novel autosomal recessive *TTN* truncating mutations: Escobar syndrome and congenital heart defect in two Brazilian patients**  
N. Linhares; A. Giannetti; D. Santos; L. Silva; S. Pena; J. Gurgel-Giannetti
- P475 **Alternative splicing in titin: new insights into exon usage**  
P. Jonson; S. Huovinen; P. Auvinen; P. Hackman; B. Udd; M. Savarese
- P476 **Muscle hypertrophy with *RYR1* mutation**  
B. Lace; P. Gould; A. Dionne; N. Chrestian
- P477 **6-Minute walk test as a fatigability measure in *RYR1*-related myopathies**  
R. Vasavada; M. Waite; I. Chrismer; M. Jain; K. Meilleur; J. Witherspoon
- P479 **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
- P480 **Establishment of an international database of titin mutations and their phenotypes – a follow up**  
M. Savarese; C. Bönnemann; A. Ferreira; 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

- P481 ***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
- P482 **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
- P483 **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
- P484 **Motor performance and disease progression in *RYR1*-RM**  
J. Witherspoon; R. Vasavada; M. Waite; I. Chrismer; M. Jain; K. Meilleur
- Management and unusual observations (P485–498)**  
*Chairpersons: Erik Niks and Jes Rahbek*
- P485 **European reference network for rare neuromuscular diseases: EURO-NMD**  
R. Leary; M. Hails; H. Lochmüller; K. Bushby Newcastle University; T. Evangelista
- P486 **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
- P487 **Drop-out in longitudinal natural history studies in neuromuscular diseases**  
D. Ho; V. Chê; A. Phelep; L. Servais; M. Annoussamy
- P488 **Using a traffic light system to aid advance care planning in neuromuscular patients**  
C. Bassie; T. Willis; R. Kulshrestha; R. Vithlani
- P489 **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
- P490 **Palliative medicine and neuromuscular disorders in adult's diseases and symptoms**  
Y. Easthope-Mowatt; C. Bassie; M. McFarlene; R. Kulshrestha; T. Willis; D. Willis
- P491 **Walking with weakness: a snapshot of gait in a paediatric neuromuscular clinic**  
K. de Valle; M. Ryan; A. Kornberg; R. Kennedy; K. Carroll
- P492 **Movement disorders hidden in the neuromuscular clinic**  
S. Paus; J. Reimann
- P493 ***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
- P494 **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
- P495 **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
- P496 **Axial myopathy: clinical and histopathological features in 7 patients**  
S. Choi; S. Ahn; S. Lim; J. Sung; J. Shin
- P497 **Axial myopathy in patients with neuromuscular diseases**  
K. Knak; N. Witting; J. Vissing; K. Rudolf
- P498 **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. Mingozi
- 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. Collella; 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. Mingozi
- TH.O.19 **Tamoxifen increases survival, improves motor function and reduces levels of BIN1 and DNMT2 in a mouse model of X-linked centronuclear (myotubular) myopathy**  
E. Gayi; H. Ismail; B. Cowling; L. Neff; J. Laporte; L. Scapozza; O. Dorchie
- 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. Wehl; 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 Topaloglu*
- 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



Oct 4 (Reuters) - Summit Therapeutics Plc : :Summit highlights DMD biomarker validation data and phase 2 clinical trial baseline characteristics in presentations at 22nd WMS Congress.Summit highlights DMD biomarker validation data and phase 2 clinical trial baseline characteristics in presentations at 22nd WMS congress.Summit Therapeutics Plc - â€œexpect to report first 24-week biopsy, MRI and functional data from phaseout DMD.