icnmd XIII

WFN
WORLD FEDERATION OF NEUROLOGY

13th INTERNATIONAL CONGRESS ON NEUROMUSCULAR DISEASES

DEDICATED TO MUSCLE, NEUROMUSCULAR JUNCTION, PERIPHERAL NERVE AND MOTOR NEURON DISEASES

PROGRAM

NICE - FRANCE
5-10 JULY 2014

ACROPOLIS CONVENTION CENTRE

www.icnmd2014.org

UNDER THE AUSPICES OF

sfm SOCIÉTÉ FRANÇAISE DE MYOLOGIE
SFNP
TREAT-NMD Neuromuscular Network
SFN Société Française de Neurologie
PNS PERIPHERAL NERVE SOCIETY
EUROPEAN NEUROMUSCULAR CENTRE

WITH THE SUPPORT FROM

Association Monégasque Contre les Myopathies
AFMTELETHON INNOVER POUR GUERIR
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CONSEIL GÉNÉRAL ALPES-MARITIMES
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icnmd XIII

Dear Colleagues,

On behalf of the Scientific and Organizing Committee of the 13th International Congress on Neuromuscular Diseases, I am delighted to welcome you in Nice in the opportunity of this major event of exchanges and circulation of knowledge for those interested in care and scientific advances in the field.

ICNMD is the regular meeting of the Research Group on Neuromuscular Diseases-World Federation of Neurology (RGNMD-WFN). The first one was organized 45 years ago, in 1969, in Milan – Italy, then in 1971, in Perth – Australia, since hosted on a 4-year event basis by several medical and research expert groups all around the world and firmly established as an important forum by the global neuromuscular community. The Organizing Committee of the 13th ICNMD is proud to have been selected by the Executive Committee of the RGNMD-WFN to manage the 2014 version and wishes to express its thanks for this commitment. I may suggest on this occasion, as the scientific production is incredibly growing in the theme, the congress would be organized every 2 years in the future.

Thanks to all attendees, ICNMD is a unique chance to share scientific advances in improving care, understanding disease pathogenesis, and developing innovative treatments in muscle, neuromuscular junction, peripheral neuropathies and motor neuron diseases. I hope we will reach your expectation in this present event, for young colleagues as well as for established expert physicians and researchers, in upgrading knowledge, and to have time for stimulating discussions and exchanges of views and ideas in your focused concerns.

I wish to thank international expert speakers who accepted to contribute in plenary lectures, workshops, breakfast seminars and courses as the attendees who carried to us their latest works. We organized the scientific program with in the mind everyone will find enough room for presenting and discuss updated data. Selected abstracts have the chance of a 3-slides oral plenary presentation.

I wish to thank the AFM-Téléthon, and other non-profit organizations, having developed for a long time special favorable conditions for the academic interest in France for neuromuscular diseases. I wish also to thank all individuals and companies who contributed to manage the "back-stage" of such an international meeting and helped us in welcoming you.

In the meantime, hosting the congress in the city of Nice is a fantastic occasion to visit the French Riviera at this time of the year and I hope you may associate hard working time and some relaxing promenades discovering cultural aspects of our splendid city and breath taking views of the vicinity.

I wish you to enjoy the congress and your visit in the capital of La Côte d'Azur.

Claude Desnuelle, MD, PhD
President of the 13th ICNMD

PROGRAM AT A GLANCE

SATURDAY JULY 5

16:00	OPENING OF THE WELCOME DESK - ONSITE REGISTRATIONS

SUNDAY JULY 6

	LEVEL 2		LEVEL 3		
	Auditorium Athena	Auditorium Hermes	Room Clio	Room Thalie	Room Erato
08:30-10:00		TC1	TC2	TC3	TC4
10:00-10:30	COFFEE BREAK				
10:30-12:00		TC5	TC6	TC7	TC8
12:00-14:00	FREE LUNCH				
14:00-15:30		TC11	TC10	TC9	TC12
15:30-16:00	COFFEE BREAK				
16:00-17:30		TC15	TC14	TC13	TC16
18:30	OPENING CEREMONY				

MONDAY JULY 7

	LEVEL 2		LEVEL 3			
	Auditorium Athena	Auditorium Hermes	Room Clio	Room Thalie	Room Erato	Room Uranie
08:00-10:00	PLENARY SESSION 1					
10:00-10:30	COFFEE BREAK					
10:30-11:30	POSTER FLASH SESSION 1					
11:30-12:30	POSTER SESSION 1					
12:30-14:30	FREE LUNCH	INDUSTRY SPONSORED SYMPOSIUM	FREE LUNCH			
14:30-16:00	THEMATIC WORKSHOP SESSION 1 - TW1.1	THEMATIC WORKSHOP SESSION 1 - TW1.5	THEMATIC WORKSHOP SESSION 1 - TW1.3	THEMATIC WORKSHOP SESSION 1 - TW1.4	THEMATIC WORKSHOP SESSION 1 - TW1.2	
16:00-16:30	COFFEE BREAK					
16:30-18:00	THEMATIC WORKSHOP SESSION 2 - TW2.6	THEMATIC WORKSHOP SESSION 2 - TW2.2	THEMATIC WORKSHOP SESSION 2 - TW2.3	THEMATIC WORKSHOP SESSION 2 - TW2.4	THEMATIC WORKSHOP SESSION 2 - TW2.5	THEMATIC WORKSHOP SESSION 2 - TW2.1

TUESDAY JULY 8

	LEVEL 2		LEVEL 3		
	Auditorium Athena	Auditorium Hermes	Room Clio	Room Thalie	Room Erato
07:00-08:00			BREAKFAST SEMINAR 1		
08:00-10:00	PLENARY SESSION 2				
10:00-10:30	COFFEE BREAK				
10:30-11:30	POSTER FLASH SESSION 2				
11:30-12:30	POSTER SESSION 2				
12:30-14:30	FREE LUNCH	INDUSTRY SPONSORED SYMPOSIUM	FREE LUNCH		
14:30-16:00	THEMATIC WORKSHOP SESSION 3 - TW3.4	THEMATIC WORKSHOP SESSION 3 - TW3.2	THEMATIC WORKSHOP SESSION 3 - TW3.3	THEMATIC WORKSHOP SESSION 3 - TW3.1	THEMATIC WORKSHOP SESSION 3 - TW3.5
16:00-16:30	COFFEE BREAK				
16:30-18:00	THEMATIC WORKSHOP SESSION 4 - TW4.4	THEMATIC WORKSHOP SESSION 4 - TW4.2	THEMATIC WORKSHOP SESSION 4 - TW4.3	THEMATIC WORKSHOP SESSION 4 - TW4.1	THEMATIC WORKSHOP SESSION 4 - TW4.5

WEDNESDAY JULY 9

	LEVEL 2		LEVEL 3			
	Auditorium Athena	Auditorium Hermes	Room Clio	Room Thalie	Room Erato	Room Calliope
07:00-08:00			BREAKFAST SEMINAR 2			
08:00-10:00	PLENARY SESSION 3					
10:00-10:30	COFFEE BREAK					
10:30-11:30	POSTER FLASH SESSION 3					
11:30-12:30	POSTER SESSION 3					
12:30-14:30	FREE LUNCH		INDUSTRY SPONSORED SYMPOSIUM	FREE LUNCH		INDUSTRY SPONSORED SYMPOSIUM
14:30-16:00	THEMATIC WORKSHOP SESSION 5 - TW5.1	THEMATIC WORKSHOP SESSION 5 - TW5.2	THEMATIC WORKSHOP SESSION 5 - TW5.3	THEMATIC WORKSHOP SESSION 5 - TW5.4	THEMATIC WORKSHOP SESSION 5 - TW5.5	
16:00-16:30	COFFEE BREAK					
16:30-18:00	THEMATIC WORKSHOP SESSION 6 - TW6.1	THEMATIC WORKSHOP SESSION 6 - TW6.2	THEMATIC WORKSHOP SESSION 6 - TW6.3	THEMATIC WORKSHOP SESSION 6 - TW6.4	THEMATIC WORKSHOP SESSION 6 - TW6.5	

THURSDAY JULY 10

	LEVEL 2		LEVEL 3			
	Auditorium Athena	Auditorium Hermes	Room Clio	Room Thalie	Room Erato	Room Calliope
07:00-08:00			BREAKFAST SEMINAR 3			
08:00-10:00	PLENARY SESSION 4					
10:00-10:30	COFFEE BREAK					
10:30-11:30	POSTER FLASH SESSION 4					
11:30-12:30	POSTER SESSION 4					
12:30-14:30	FREE LUNCH					INDUSTRY SPONSORED SYMPOSIUM
14:30-16:00	THEMATIC WORKSHOP SESSION 7 - TW7.1	THEMATIC WORKSHOP SESSION 7 - TW7.3	THEMATIC WORKSHOP SESSION 7 - TW7.2	THEMATIC WORKSHOP SESSION 7 - TW7.4	THEMATIC WORKSHOP SESSION 7 - TW7.5	
16:00-16:30	COFFEE BREAK					
16:30-18:00	THEMATIC WORKSHOP SESSION 8 - TW8.3	THEMATIC WORKSHOP SESSION 8 - TW8.2	THEMATIC WORKSHOP SESSION 8 - TW8.1	THEMATIC WORKSHOP SESSION 8 - TW8.4		
18:30-19:00	CLOSING CEREMONY					

08:30-10:00

AUDITORIUM HERMES **TC1** **Arthrogryposis and other muscle diseases with prominent contractures: new and main entities, diagnostic strategies.**

- Muscle diseases with prominent joint contractures: main entities and diagnostic strategy. *Bruno Eymard (Paris, France - Coordinator)*
- Arthrogryposis multiplex congenita: new and main entities, diagnostic strategies. *Judith Melki (Paris, France)*

ROOM CLIO **TC2** **Analysis of muscle tissue with electron microscopy in research practice.**
Clara Franzini-Armstrong (Philadelphia, USA - Coordinator)

ROOM THALIE **TC3** **Muscle biopsy in diagnosing neuromuscular weakness, peripheral sensory loss, and some pain syndromes and s-IBM.**

- Value of muscle biopsy in diagnosing neuromuscular weakness, peripheral-nerve sensory loss and some pain syndromes. *William K Engel (Los Angeles, USA - Coordinator)*
- Revival of diagnostic electron microscopy in neuromuscular pathology. *Michel Fardeau (Paris, France)*
- Pathologic diagnostic criteria of s-IBM muscle biopsy. *Valerie Askanas (Los Angeles, USA)*

ROOM ERATO **TC4** **Mitochondrial medicine: conundrums, challenges and hopes.**

- Coenzyme Q Deficiency in Muscle. A Supercomplex Issue. *Leonardo Salviati (Padua, Italy - Coordinator)*
- The neuromuscular target in mitochondrial disorders. *Antonio Toscano (Messina, Italy)*
- Mitochondrial disorders, from modern genetics to experimental therapy. *Massimo Zeviani (Cambridge, UK)*

10:00-10:30 COFFEE BREAK

10:30-12:00

AUDITORIUM HERMES **TC5** **Periodic paralysis and the calcium channel.**

- Non dystrophic myotonias. *Giovanni Meola (Milan, Italy - Coordinator)*
- Periodic paralysis and calcium channel. *Bertrand Fontaine (Paris, France)*

ROOM CLIO **TC6** **Electrophysiologic approaches of acute and chronic polyradiculoneuropathies.**

- CIDP : update on clinical features, variants and electrophysiological studies. *Shahram Attarian (Marseille, France - Coordinator)*
- Electrophysiological and immunopathological correlates of GBS subtypes. *Antonino Uncini (Chieti, Italy)*

ROOM THALIE **TC7** **How to explore patients when using new genetic Tools.**

- How to explore patients when using new genetic Tools. *Vincenzo Nigro (Naples, Italy - Coordinator)*
- Diagnostic possibilities offered by new genetic technologies in neuromuscular disorders. *Marc Bartoli (Marseille, France)*

ROOM ERATO **TC8** **Congenital myasthenic syndromes.**

- Basic aspects of congenital myasthenic syndromes. *Daniel Hantaï (Paris, France - Coordinator)*
- Clinical presentation of congenital myasthenic syndromes Bruno Eymard. *Bruno Eymard (Paris, France)*

12:00-14:00 FREE LUNCH

14:00-15:30

ROOM THALIE

TC9 New concepts in Facioscapulohumeral Muscular Dystrophy.

- Facioscapulohumeral muscular dystrophy type 1, clinical features and symptomatic treatments. *Baziel van Engelen (Nijmegen, The Netherlands)*
- If not FSHD1, what else could this be? Differential diagnosis with FSHD2 and others FSHD mimicking conditions. *Sabrina Sacconi (Nice, France - Coordinator)*
- Genetic, epigenetic and new therapeutic strategies for FSHD. *Sylvère Van der Maarel (Leiden, The Netherlands)*

ROOM CLIO

TC10 Amyloid neuropathy: diagnostic tools and treatments.

- Amyloid neuropathy: diagnostic tools and treatments. *David Adams (Le Kremlin Bicêtre, France - Coordinator)*
- Diagnostic hallmarks and pitfalls in late-onset progressive transthyretin-related amyloid-neuropathy. *Kristl Claeys (Aachen, Germany)*

AUDITORIUM HERMES

TC11 Basic concepts in the cellular biology of muscle: from neuromuscular transmission to contraction.

- Neuromuscular transmission. *Laurent Schaeffer (Lyon, France)*
- From muscle excitation to contraction. *Bruno Allard (Lyon, France - Coordinator)*

ROOM ERATO

TC12 SFM teaching course: Discussion of clinical cases of myopathy.

- *Françoise Chapon (Caen, France - Coordinator)*
- *Andoni Urtizberea (Paris, France)*

15:30-16:00 COFFEE BREAK

16:00-17:30

ROOM THALIE

TC13 What about the classification in Limb Girdle Muscular Dystrophy?

- What about the classification in LGMD. *Corrado Angelini (Padua, Italy - Coordinator)*
- Genetic basis of limb-girdle muscular dystrophies: the 2014 update. *Vincenzo Nigro (Naples, Italy)*

ROOM CLIO

TC14 Care and treatment in ALS: new developments.

- Therapy in ALS: problems and prospects. *Vincenzo Silani (Milan, Italy – Coordinator)*
- Therapies in ALS/MND. New developments in the symptomatic management of ALS/MND. *Pamela Shaw (Sheffield, United Kingdom)*
- Genetic-Based Antisense Oligonucleotide Therapy in ALS. *John Landers (Boston, USA)*

AUDITORIUM HERMES

TC15 How to use imaging techniques in the diagnostic of myopathies.

- Use of MRI in clinical practice in the diagnosis of myopathies. *Anthony Behin (Paris, France - Coordinator)*
- Muscle MRI and myopathies. *Robert Carlier (Garches, France)*

ROOM ERATO

TC16 New insights in muscle glycogenoses diseases

- Glycogen depletion and polyglucosan storage myopathies. *Anders Oldfors (Gothenburg, Swenden - Coordinator)*
- Muscle glycogenoses: different phenotypes and pathophysiology. *John Vissing (Copenhagen, Denmark)*
- Pompe Disease: a "sui generis" disorder. *Antonio Toscano (Messina, Italy)*

08:00 **PLENARY LECTURE SESSION 1** (AUDITORIUM ATHENA)

BASIC SCIENCES, MUSCLE AND NERVE DEVELOPMENT

Chairpersons: Michael Rudnicki (Ottawa, Canada), Ketan Patel (Reading, United Kingdom)

PL1.1 **Modelling Duchenne Dystrophy with embryonic stem cells.** *Olivier Pourquié (Strasbourg, France)*
PL1.2 **Regulation of Muscle Satellite Cells.** *Margaret Buckingham (Paris, France)*
PL1.3 **Novel signalling pathways that control muscle mass.** *Marco Sandri (Padua, Italy)*
PL1.4 **Genetic and epigenetic determinants of myogenesis.** *Stephen Tapscott (Seattle, USA)*

10:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

10:30 **POSTER FLASH SESSION 1** (AUDITORIUM ATHENA)

Moderators: Gisèle Bonne (Paris, France), Pascal Maire (Paris, France)

PS1 -1/#138 Role of Serum response factor in muscular satellite cells. *Voahangy Randrianarison-Huetz, Aikaterini Papaefthymiou, Laura Collard, Ulduz Faradova, Athanassia Sotiropoulos (Paris, France)*

PS1-23/#464 Novel gene networks and biological pathways regulated by FoxO in skeletal muscle during cancer cachexia. *Sarah Judge, Brandon Roberts, Chia-Ling Wu, Adam Beharry, Leonardo Ferreira, Susan Kandarian, Andrew Judge (Gainesville, USA)*

PS1-13/#379 Cytoskeleton network regulation and nuclear positioning during muscle development.
Bruno Cadot, Edgar Gomes, Vincent Gache (Paris, France)

PS1-27/#406 Characterization of the trafficking and functional properties of the muscle specific long STIM1 isoform.
Laurent Bernheim, Pierre Cosson, Nicolas Demaurex, Maud Frieden, Sophie Sauc (Geneva, Switzerland)

PS1-5/#487 Muscle Ring Finger-1 (MuRF1) activity is present in cardiac mitochondria and regulates reactive oxygen species production in vivo.
Taylor Mattox, Martin Young, Mathias Gautel, Ethan Anderson, Monte Willis (Greenville, USA)

PS1-25/#258 Defective mechanosensing responses are associated with altered Yes-associated protein (YAP) signaling in myoblasts from human muscular dystrophies. *Tsolere Arakelian, Hélène Duchemin, Anne T Bertrand, Kamel Mamchaoui, Anne Bigot, Simindokht Ziaei, Thomas Voit, Gisèle Bonne, Catherine Coirault, Martina Fischer (Paris, France)*

PS2-207/#197 MRI validation of a transcriptional cascade propagation model in FSHD muscular dystrophy.
NBM Voet, AC Geurts, GW Padberg, BGM van Engelen, A Heerschap, BH Janssen (Nijmegen, Netherlands)

PS2-152/#292 Reducing dynamin 2 rescues myotubular myopathy in mice. *Belinda Cowling, Thierry Chevremont, Ivana Prokic, Christine Kretz, Arnaud Ferry, Catherine Coirault, Vincent Laugel, Norma Romero, Jocelyn Laporte (Illkirch, France)*

PS2-171/#106 The pathogenesis of desminopathies: lessons from R350P desmin knock-in mice. *Christoph Clemen, Florian Stöckigt, Frederic Chevessier, Karl-Heinz Strucksberg, Lilli Winter, Harald Herrmann, Matthias Türk, Regine Schneider-Stock, Oliver Friedrich, Rainer Meyer, Oliver Müller, Jan Wilko Schrickel, Rolf Schröder (Cologne, Germany)*

PS1-24/#486 Mrtf/Srf are mediators of mechanotransduction in skeletal muscle. *Laura Collard, Lorraine Montel, Sylvie Hénon, Athanassia Sotiropoulos, Voahangy Randrianarison, Alessandra Pincini (Paris, France)*

PS1-126/#356 Full-Length Dysferlin Expression Driven by Engineered Human Dystrophic Blood-Derived CD133+ Stem Cells.
Mirella Meregalli, Clementina Sitzia, Claire Navarro, Andrea Farini, Erica Montani, Nicolas Wein, Paola Razini, Cyriaque Beley, Letizia Cassinelli, Marzia Belicchi, Dario Parazzoli, Luis Garcia, Yvan Torrente (Milan, Italy)

PS1-20/#107 The effect of aerobic training and detraining on mithochondrial function in aging. *Frank Thøgersen, Marie Loui Sveen, Mette Cath Ørngreen, Thomas Krag, John Vissing, Tina Dysga Jeppesen, Søren Pete Andersen (Copenhagen, Denmark)*

PS1-37/#84 The Wnt receptor Frizzled 3 is required for nerve-muscle target recognition process during neuromuscular junction formation.
Julien Messéant, Perrine Delers, Claire Legay, Laure Strochlic (Paris, France)

PS3-323/#519 Molecular mechanisms of RAPSN mutations in congenital myasthenic syndromes.
David Beeson, Judith Cossins, Jacqueline Palace, Jonathan Cheung Oxford (United Kingdom)

PS1-42/#422 Neuromuscular junction integrity is dependent on rapsyn/AChR complex-intermediate filament linkage via plectin.
Marianne Raith, Ruth Herbst, Gerhard Wiche, Eva Mihailovska (Vienna, Austria)

PS3-303/#131 In vitro characterization of satellite cells from myasthenic patients. *Marie Maurer, Kamel Mamchaoui, Yoan Bismuth, Sylvain Bourgoin, Vincent Mouly, Gillian Butler-Browne, Sonia Berrih-Aknin, Mohamed Attia (Paris, France)*

PS3-285/#165 Phosphorylation of the autophagy receptor NBR1 by GSK3 modulates protein aggregation and is abnormal in muscles of sporadic inclusion body myositis patients. *Francesca Lo Verso, Francesca Ratti, Fanny Pilot-Storck, Nathalie Streichenberger, Marco Sandri, Laurent Schaeffer, Evelyne Goillot, Anne-Sophie Nicot (Lyon, France)*

PS4-393/#146 Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. *Laura Ferraiuolo, Carlos Miranda, Shibi Likhite, Arthur Burghes, Stephen Kolb, Brian Kaspar, Kathrin Meyer (Columbus, USA)*

PS4-415/#59 The pathogenic contribution of astrocytic and muscular TWEAK to ALS pathology. *Melissa Bowerman, Céline Salsac, Emmanuelle Coque, Frédérique Scamps, Alexandre Brodovitch, Cedric Raoul (Montpellier, France)*

11:30 **POSTER SESSION 1** *(Authors on allocated panel - See Poster sessions list on page 21)*

VISIT OF THE EXHIBITION

12:30 **INDUSTRY SPONSORED SYMPOSIUM** (AUDITORIUM HERMES)

14:30 **THEMATIC WORKSHOPS - SESSION 1**

TW 1.1 DEVELOPMENT OF NEUROMUSCULAR JUNCTION (AUDITORIUM ATHENA)

Coordinator: Laurent Schaeffer (Lyon, France)

1.1.1 **Regulation of muscle gene expression by electrical activity: CtBP mediates repression.** *Laurent Schaeffer (Lyon, France)*
1.1.2 **Making and breaking neuromuscular synapses.** *Steven Burden (New York, USA)*
1.1.3 **Signaling perturbations at the NMJ.** *David Glass (Boston, USA)*

TW 1.2 VASCULITIC PERIPHERAL NEUROPATHY (ROOM ERATO)

Coordinator: Michael P Collins (Milwaukee, USA)

1.2.1 **Classification and pathology of the systemic and non systemic vasculitic neuropathies.** *Michael P Collins (Milwaukee, USA)*
1.2.2 **Clinical approach to vasculitic neuropathies.** *Alexander FJE Vrancken (Utrecht, The Netherlands)*
1.2.3 **Treatment of vasculitic neuropathies.** *Robert DM Hadden (London, United Kingdom)*

TW 1.3 SPINAL MUSCULAR ATROPHY (ROOM CLIO)

Coordinator: Arthur Burghes (Columbus, USA)

1.3.1 **Development of therapies for SMA preclinical.** *Arthur Burghes (Columbus, USA)*
1.3.2 **Dysregulation of RNA processing in spinal muscular atrophy.** *Livio Pellizzoni (New York, USA)*
1.3.3 **Clinical trials and measures in SMA.** *Eugenio Mercuri (Rome, Italy)*

TW 1.4 STEM CELLS AND MUSCLE REGENERATION (ROOM THALIE)

Coordinator: Gillian Butler-Browne (Paris, France)

1.4.1 **Regulation of satellite cell migration manipulating the amoeboid mechanisms for cell movement to promote skeletal muscle regeneration.** *Ketan Patel (Reading, United Kingdom)*
1.4.2 **Eph/ephrin interactions generate fiber type specificity during muscle development, regeneration and reinnervation.** *Dawn Cornelison (Columbia, USA)*
1.4.3 **Wnt signaling(s) and skeletal muscle regeneration.** *Fabien Le Grand (Paris, France)*

TW 1.5 INCLUSION BODY MYOSITIS (AUDITORIUM HERMES)

Coordinator: Valerie Askanas (Los Angeles, USA)

1.5.1 **Critical pathogenic mechanisms associated with the muscle-fiber degeneration specific to Sporadic Inclusion-Body Myositis (s-IBM), and their treatment possibilities.** *Valerie Askanas (Los Angeles, USA)*
1.5.2 **How the impaired muscle repair and regeneration contribute to the sIBM pathogenesis, and their treatment possibilities.** *Massimiliano Mirabella (Rome, Italy)*
1.5.3 **How do immune-mediated mechanisms contribute to sIBM pathogenesis, and why dysimmune treatments are largely not effective?** *Marinos Dalakas (Athens, Greece / Philadelphia, USA)*

16:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

16:30 **THEMATIC WORKSHOPS - SESSION 2**

TW 2.1 DEVELOPMENT OF MUSCLE LINEAGES (HEART/SKELETAL) (ROOM URANIE)

Coordinator: Christophe Marcelle (Melbourne, Australia)

2.1.1 **Morphogenesis of muscles: a bird'eye view.** *Christophe Marcelle (Melbourne, Australia)*
2.1.2 **Head muscle origin, development and regeneration.** *Eldad Tzahor (Rehovot, Israel)*
2.1.3 **Regulation of tissue-specific alternative splicing during development of muscle lineage.** *Thomas Braun (Bad Nauheien, Germany)*

TW 2.2 DYSGLOBULINEMIC AND LYMPHOMA-RELATED PERIPHERAL NEUROPATHIES (AUDITORIUM HERMES)

Coordinator: Thierry Kuntzer (Lausanne, Switzerland)

2.2.1 **Dysglobulinemic neuropathies.** *Thierry Kuntzer (Lausanne, Switzerland)*
2.2.2 **Lymphoma associated neuropathies.** *Wolfgang Grisold (Vienna, Austria)*
2.2.3 **Treatment options in haemopathic-associated neuropathies.** *Marinos Dalakas (Athens, Greece / Philadelphia, USA)*

TW 2.3 PATHOMECHANISMS OF HEREDITARY NEUROPATHIES (ROOM CLIO)

Coordinator: Vincent Timmerman (Antwerp, Belgium)

2.3.1 **Understanding the pathomechanisms of peripheral nerve amyloid.** *Mary M Reilly (London, United Kingdom)*
2.3.2 **Understanding the pathomechanisms of hereditary sensory and autonomic neuropathies.** *Ingo Kurth (Jena, Germany)*
2.3.3 **Understanding the pathomechanisms of distal hereditary motor neuropathies.** *Vincent Timmerman (Antwerpen, Belgium)*

TW 2.4 CALCIUM FLUXES AND ION CHANNELS IN MUSCLE (ROOM THALIE)

Coordinator: Robert T Dirksen (Rochester, NY, USA)

2.4.1 **Periodic Paralysis related to calcium channelopathy.** *Bertrand Fontaine (Paris, France)*
2.4.2 **Genetic evidence that Ca2+ is the key inducer of myofiber necrosis in muscular dystrophy.** *Jeffery D Molkentin (Cincinnati, USA)*
2.4.3 **Identification of a "Relay Gate" in the type I ryanodine receptor from functional analyses of a cluster of disease mutations in TM2.** *Robert T Dirksen (Rochester, NY, USA)*

TW 2.5 MYOFIBRILLAR AND GNE MYOPATHIES (ROOM ERATO)

Coordinator: Bjarne Udd (Tampere, Finland)

2.5.1 **Myofibrillar myopathies - clinical characteristics.** *Bjarne Udd (Tampere, Finland)*
2.5.2 **Diagnostic clues from muscle pathology and MRI in Myofibrillar Myopathies.** *Montse Olivé (Barcelona, Spain)*
2.5.3 **Protein aggregates and Rimmed Vacuoles: Possible therapeutic interventions in MFM.** *Conrad Weihl (St louis, USA)*

TW 2.6 ENMC SESSION: HIGHLIGHTS FROM RECENT ENMC WORKSHOPS (AUDITORIUM ATHENA)

Coordinator: Baziel van Engelen, Alexandra Breukel (Nijmegen, The Netherlands)

2.6.1 **Care for adults with DMD – outcome of an ENMC workshop.** *Jes Rahbek (Aarhus, Denmark)*
2.6.2 **Biomarkers in DMD: from the discovery to the development toward clinical application and translation in other NMDs.** *Alessandra Ferlini (Ferrara, Italy)*
2.6.3 **Autophagy in Muscular Dystrophies: Translational approach.** *Luciano Merlini (Bologna, Italy)*

07:00 **BREAKFAST SEMINAR 1** (ROOM CLIO)

New insights into skeletal muscle membrane function. *Elizabeth McNally (Chicago, USA)*

08:00 **PLENARY LECTURE SESSION 2** (AUDITORIUM ATHENA)

GENETICS IN NEUROMUSCULAR DISEASES

Chairpersons: Sylvère van der Maarel (Leiden, The Netherlands), Vincenzo Nigro (Naples, Italy)

PL 2.1 **Genetics of dystroglycanopathies: new advances in dystroglycan post-translational processing.** *Kevin Campbell (Iowa City, USA)*

PL 2.2 **Single gene, gene panel and exome sequencing approaches for neuromuscular diseases.** *Nicolas Lévy (Marseille, France), Madhuri Hegde (Atlanta, USA)*

PL 2.3 **Congenital Myopathies: The New Genetic Era.** *Carsten G Bönnemann (Bethesda, USA)*

PL 2.4 **New genes in motor neuron diseases.** *Wim Robberecht (Leuven, Belgium)*

10:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

10:30 **POSTER FLASH SESSION 2** (AUDITORIUM ATHENA)

Moderators: Isabelle Marty (Grenoble, France), Nicolas Lévy (Marseille, France)

PS2-136/#192 Impaired viability of muscle precursor cells in muscular dystrophy with glycosylation defects and amelioration of its severe phenotype by limited gene expression. *Chih-Chieh Yu, Chiyomi Ito, So-ichiro Fukada, Tomoko Chiyo, Kazuhiro Kobayashi, Takashi Okada, Shin'ichi Takeda, Tatsushi Toda, Motoi Kanagawa (Kobe, Japan)*

PS2-150/#203 Multimodal MRI and 31P-MRS investigations of the ACTA1(Asp286Gly) mouse model of nemaline myopathy provide evidence of impaired in vivo muscle function, altered muscle structure and disturbed energy metabolism. *Charlotte Gineste, Guillaume Duhamel, Yann Le Fur, Christophe Vilmen, Patrick J. Cozzone, Dav/ #Bendahan, Julien Gondin (Marseille, France)*

PS2-180/#451 ZASP mutations in actin-binding domain cause disruption of skeletal muscle actin filaments in myofibrillar myopathy. *Xiaoyan Lin, Janelle Ruiz, Ilda Bajraktari, Rachel Ohman, Kenneth Fischbeck, Robert Griggs, Ami Mankodi (Bethesda, USA)*

PS2-204/#497 Digenic expression of sodium and chloride channel mutations in patients with non dystrophic myotonia. *Alain Furby, Savine Vicart, Jean-Philippe Camdessanche, Emmanuel Fournier, Stéphane Chabrier, Emmanuelle Lagrue, Renaud Touraine, Bertrand Fontaine, Damien Sternberg (Saint-Etienne, France)*

PS2-265/#181 Two founder mutations within GNE gene and high prevalence of GNE myopathy identified in North of Britain. *Amina Chaouch, Kathryn Brennan, Judith Hudson, Cheryl Longman, John McConville, Patrick Morrison, Maria Farrugia, Richard Petty, Willie Stewart, Fiona Norwood, Rita Horvath, Patrick Chinnery, Donald Costigan, John Winer, Tuomo Polvikoski, Estelle Healy, Anna Sarkozy, Michela Guglieri, Teresinha Evangelista, Michelle Eagle, Kate Bushby, Volker Straub, Hanns Lochmüller, Oksana Pogoryelova (Newcastle Upon Tyne, United Kingdom)*

PS2-268/#429 Analysis of baseline sialic acid and NCAM data in GNE myopathy patients & mouse model. *Paul Lee, Jaclyn Cadaoas, Gabrielle Morris, Emil Kakkis, Michel Vellard, Yiumo Chan (Novato, USA)*

PS4-455/#308 Targeted and exome sequencing for diagnosis and novel gene identification in congenital myopathies. *Valerie Biancalana, Johann Bohm, Osorio Lopes Abath Neto, Edoardo Malfatti, Nicolas Dondaine, Nasim Vasli, Norma Romero, Jocelyn Laporte (Illkirch, France)*

PS1-99/#551 Comparison of rAAV6 and rAAV9 transduction in canines via jugular vein infusion. *Jane Seto, Julian Ramos, Stephen Hauschk, Guy Odom, Jeffrey Chamberlain (Seattle, USA)*

PS2-138/#346 Novel collagen VI chains in zebrafish skeletal muscle. *Claire Rocancourt, Jeanne Lainé, Arnaud Klein, Laura Lyphout, Corine Gartioux, Edor Kabashi, Xavier Cousin, Valérie Allamand, Laetitia Ramanoudjame (Paris, France)*

PS2-226/#85 Pompe disease: pathophysiology and novel approaches to therapy. *Nina Raben (Bethesda, USA)*

PS1-39/#215 Integrins are required for synaptic transmission and development of the neuromuscular junction. *Jacob Ross, Richard Webster, Tanguy Lechertier, Francesco Muntoni, Jennifer Morgan, Kairbaan Hodivala-Dilke, Francesco Conti (London, UK)*

PS1-41/#326 The Molecular Machinery that Orchestrates Neuromuscular Junction Remodeling. *Pawe Niewiadomski, Joshua Sanes, Tomasz Prószyski (Warsaw, Poland)*

PS2-252/#467 Pathogenesis of the neuromuscular junction and motor neuron in Pompe disease. *A. Gary Todd, Sooyeon Lee, David Fuller, Lucia Notterpek, Barry Byrne, Darin Falk (Gainesville, USA)*

PS3-273/#261 Redifining the immune histolochemical pattern of Anti-SRP auto-antibody positive patients: a subgroup present significant inflammation. *Aude Rigolet, Tanya Stojkovic, Pascal Laforet, Antony Behin, Bruno Eymard, Norman Zerbe, Peter Hufnagl, Thierry Maisonobe, Kuberaka Mariampillai, Corinna Preusse, Serge Herson, Olivier Benveniste, Werner Stenzel , Yves Allenbach (Berlin, Germany)*

PS3-274/#264 Myofiber HLA-DR expression is distinctive biomarker for antisynthetase myositis. *Jessie Aouizerate, Marie De Antonio, Thierry Maisonobe, Yasmine Baba-Amer, Romain K Gherardi, Francis Berenbaum, Loic Guillevin, Olivier Benveniste, Francois Jerome Authier (Creteil, France)*

PS4-420/#172 Muscle mitochondrial dysfunction in a large cohort of genetically-determined SMA patients. *Michela Ripolone, Dario Ronchi, Raffaella Violano, Dionis Vallejo, Emanuele Barca, Gigliola Fagiolari, Angela Berardinelli, Umberto Ballottin, Lucia Morandi, Marina Mora, Andreina Bordoni, Francesco Fortunato, Antonio Toscano, Monica Sciacco, Salvatore Di Mauro, Giacomo Pi Comi, Maurizio Moggio (Milano, Italy)*

PS3-358/#229 Improper mitochondrial calcium homeostasis is responsible for the Friedreich ataxia neural pathophysiology. *Belén Mollá, Diana Carolina Muñoz Lasso, Francesc Palau, Pilar Gonzalez-Cabo (Valencia, Spain)*

PS4-405/#558 ALS astrocytes kill motor neurons via ligation of death receptor 6 by a fragment of N-APP/APLP1. *Virginia Le Verche, Burcin Ikiz, Mariano Alvarez, Kristin Politi, Paschalis-Toma Doulias, Dimitra Papadimitriou, Todd Greco, Anatoly Nikolaiev, Andrea Califano, Harry Ishiropoulos, Manuel Than, Marc Tessier-Lavigne, Serge Przedborski, Diane B. Re (New-York, USA)*

PS3-336/#238 Gene Expression Changes in Chronic Inflammatory Demyelinating Polyneuropathy Skin Biopsies. *Andreas Steck, Adrian Panaite, Stefania Puttini, Nicolas Mermod, Susanne Renaud, Thierry Kuntzer (Lausanne, Switzerland)*

11:30 **POSTER SESSION 2** *(Authors on allocated panel - See Poster sessions list on page 25)*

VISIT OF THE EXHIBITION

12:30 **INDUSTRY SPONSORED SYMPOSIUM** (AUDITORIUM HERMES)

14:30 **THEMATIC WORKSHOPS - SESSION 3**

TW 3.1 STEM CELLS AND MUSCLE REGENERATION (ROOM THALIE)

Coordinator: Michael Rudnicki (Ottawa, Canada)

3.1.1 **A cell-autonomous loss of muscle stem cell self renewal in aged muscle.** *Bradley Olwin (Boulder, USA)*
3.1.2 **The Role of the Muscle Microenvironment in Cancer-Induced Cachexia.** *Denis Guttridge (Columbus, USA)*
3.1.3 **The role of the muscle stem cell niche in regulating regenerative myogenesis.** *Michael Rudnicki (Ottawa, Canada)*

TW 3.2 CONGENITAL MYASTHENIC SYNDROMES (AUDITORIUM HERMES)

Coordinator: Daniel Hantaï (Paris, France)

3.2.1 **An emerging subgroup of congenital myasthenic syndromes due to mutations in genes associated with N-linked glycosylation.** *David Beeson (Oxford, United Kingdom)*
3.2.2 **Diagnosis and therapy of congenital myasthenic syndromes – clinical update.** *Hanns Lochmüller (Newcastle, United Kingdom)*
3.2.3 **Agrin and MuSK in congenital myasthenia.** *Daniel Hantaï (Paris, France)*
3.2.4 **Observations in recently identified congenital myasthenic syndromes (CMS).** *Andrew G Engel (Rochester, USA)*

TW 3.3 ELECTROPHYSIOLOGICAL APPROACHES IN DYSIMMUNE NEUROMUSCULAR DISEASES (ROOM CLIO)

Coordinator: Peter Van den Bergh (Brussels, Belgium)

3.3.1 **Chronic inflammatory demyelinating polyradiculoneuropathy (CIDP): electrodiagnostic evidence.** *Peter Van den Bergh (Brussels, Belgium)*
3.3.2 **Guillan-Barré syndrome: electrodiagnostic issues and challenges.** *Antonino Uncini (Chieti, Italy)*
3.3.3 **Neuromuscular junction disorders: electrodiagnostic methods.** *J Gert van Dijk (Leiden, The Netherlands)*

TW 3.4 NEW APPROACHES OF IDENTIFICATION OF GENE: NEXT GENERATION SEQUENCING (AUDITORIUM ATHENA)

Coordinator: Nicolas Lévy (Marseille, France)

3.4.1 **New systems to interprete data from NGS analysis.** *Christophe Beroud (Marseille, France)*
3.4.2 **User-driven dynamic creation of patient registries: a modular approach to bioinformatics analysis for neuromuscular diseases.** *Matthew Bellgard (Perth, Australia)*
3.4.3 **FORGE Canada Consortium: outcomes of a 2-year National Rare Disease Gene Discovey Project.** *Jacques Michaud (Montreal, Canada)*

TW 3.5 THERAPEUTIC APPROACHES IN FACIOSCAPULOHUMERAL DYSTROPHY (ROOM ERATO)

Coordinator: Sylvère van der Maarel (Leiden, The Netherlands)

3.5.1 **Molecular mechanisms in FSHD: identification of druggable targets.** *Sylvère van der Maarel (Leiden, The Netherlands)*
3.5.2 **Aerobic exercise and cognitive behavior therapy reduce fatigue and slow progression of muscle MR fatty infiltration in FSHD.** *Baziel van Engelen (Nijmegen, The Netherlands)*
3.5.3 **Biomarkers and trial readiness in FSHD1 and FSHD2; are we ready for the future?** *Sabrina Sacconi (Nice, France)*

16:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

16:30 **THEMATIC WORKSHOPS - SESSION 4**

TW 4.1 NEW INSIGHT INTO THE MECHANISM OF MUSCLE ATROPHY (ROOM THALIE)

Coordinator: David Glass (Boston, USA)

4.1.1 **New insights into the molecular mechanisms of muscle atrophy.** *Shenhav Cohen (Haifa, Israel)*
4.1.2 **FoxO, autophagia and mitochondria: the crossroad for the control of muscle.** *Marco Sandri (Padua, Italy)*
4.1.3 **Signalling mechanisms mediating muscle homeostasis.** *David Glass (Boston, USA)*

TW 4.2 INFLAMMATORY MYOPATHIES (AUDITORIUM HERMES)

Coordinator: Isabel Illa (Barcelona, Spain)

4.2.1 **Specific auto-antibodies in inflammatory myopathies: identification of new clinico pathological syndromes?** *Olivier Benveniste (Paris, Fance)*
4.2.2 **Role of innate immunity in dermatomyositis.** *Isabel Illa (Barcelona, Spain)*
4.2.3 **Treatment of inflammatory myopathies.** *Marinos Dalakas (Athens, Greece / Philadelphia, USA)*

TW 4.3 THERAPEUTIC APPROACHES TO HEREDITARY MYOPATHIES (ROOM CLIO)

Coordinator: George Dickson (London, United Kingdom)

4.3.1 **Advanced exon skipping antisense strategies for Duchenne Muscular Dystrophy.** *Matthew Wood (Oxford, United Kingdom)*
4.3.2 **Autologous myoblast cell therapy for oculopharyngeal muscular dystrophy (OPMD): a phase I/IIa clinical study.** *Capucine Trollet (Paris, France)*
4.3.3 **AAV microdystrophin gene therapy for Duchenne Muscular Dystrophy.** *George Dickson (London, United Kingdom)*

TW 4.4 CONGENITAL MYOPATHIES (AUDITORIUM ATHENA)

Coordinator: Nigel G Laing (Perth, Australia)

4.4.1 **Nemaline Myopathy and related diseases: genetics, diagnosis and therapy.** *Nigel G Laing (Perth, Australia)*
4.4.2 **Gene replacement therapy for myotubular myopathy.** *Anna Buj-Bello (Evry, France)*
4.4.3 **Understanding - and improving - sarcomeric function in Nemaline Myopathy.** *Coen Ottenheijm (Amsterdam, The Netherlands)*

TW 4.5 THERAPEUTIC APPROACH IN MYOTONIC DYSTROPHY (ROOM ERATO)

Coordinator: Denis Furling (Paris, France)

4.5.1 **Systemic delivery of a peptide-PMO of CAG sequence neutralizes mutant RNA toxicity in a mouse model of DM1.** *Andrew Leger (Boston, USA)*
4.5.2 **Systemic delivery of an antisense oligonucleotide (ASO) targeting DMPK RNA improves the phenotype of DMSXL mice.** *Dominic Jauvin (Quebec, Canada)*
4.5.3 **Targeting nuclear expanded repeats to correct RNA gain-of-function effects in DM1.** *Denis Furling (Paris, France)*

07:00 **BREAKFAST SEMINAR 2** (ROOM CLIO)

Dysferlin: roles and implications for skeletal muscle metabolism. *Miranda Grounds (Perth, Australia)*

08:00 **PLENARY LECTURE SESSION 3** (AUDITORIUM ATHENA)

IMMUNE-MEDIATED CONDITIONS IN NEUROMUSCULAR DISEASES

Chairpersons: Valérie Askanas (Los Angeles, USA), Andrew G Engel (Rochester, USA)

PL 3.1 **Targets of immunotherapy in autoimmune neuromuscular disorders and emerging new therapies.** *Marinos Dalakas (Athens, Greece / Philadelphia, USA)*

PL 3.2 **Emerging new concepts in pathogenesis of inflammatory myopathies.** *Olivier Benveniste (Paris, France)*

PL 3.3 **Emerging new concepts in pathogenesis of peripheral neuropathy.** *Hugh Willison (Glasgow, United Kingdom)*

PL 3.4 **Emerging new concepts in pathogenesis of autoimmune channelopathies.** *Angela Vincent (Oxford, United Kingdom)*

10:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

10:30 **POSTER FLASH SESSION 3** (AUDITORIUM ATHENA)

Moderators: Jean-Marc Leger (Paris, France), Bruno Eymard (Paris, France)

PS4-439/#361 Upper limb muscle fat-water quantification MRI and clinical functional evaluation in non-ambulant Duchenne muscular dystrophy. *Matthew Evans, Christopher Sinclair, Jasper Morrow, Jordan Butler, Robert Janiczek, Michael Hanna, Paul Matthews, Tarek Yousry, Francesco Muntoni , John Thornton, Valeria Ricotti (London, UK)*

PS4-417/#124 Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. *Annie Laquérriere, Jérome Maluenda, Adrien Camus, Laura Fontenas, Klaus Dieterich, Flora Nolent, Jié Zhou, Nicole Monnier, Philippe Latour, Joel Lunardi, Monica Bayes, Pierre S Jouk, Damien Sternberg, Josiane Warszawski, Ivo Gut, Marie Gonzales, Marcel Tawk, Judith Melki (Rouen, France)*

PS2-144/#70 Muscle histopathology in nebulin-related nemaline myopathy: ultrastrastructural findings correlated to disease severity and genotype. *Edoardo Malfatti, Vilma-Lott Lehtokari, Johann Böhm, Josine De Winter, Ursula Schaffer, Brigitte Estournet, Susana Quijano-Roy, Soledad Monges, Fabiana Lubieniecki, Rémi Bellance, Mai Thao Viou, Angeline Madelaine, Bin Wu, Analia Taratuto, Bruno Eymard, Katarina Pelin, Michel Fardeau, Coen Ottenheijm, Carina Wallgren-Pettersson, Jocelyn Laporte, Norma B Romero (Paris, France)*

PS2-18/#92 Cardiac involvement in myotonic dystrophy: a nationwide cohort study. *Lars Diaz, Matthis Ranthe, Helle Petri, Morten Duno, Inger Juncker, Hans Eiberg, John Vissing, Henning Bundgaard, Jan Wohlfahrt, Mads Melbye, Marie Lund (Copenhagen, Denmark)*

PS2-231/#151 A nationwide survey of Danon disease in Japan. *Hirofumi Komaki, Nobuyuki Eura, Ikuya Nonaka, Satoshi Ueno, Ichizo Nishino, Kazuma Sugie (Nara, Japan)*

PS2-212/#244 Facioscapulohumeral muscular dystrophy type 1: quantitative MR imaging and clinical correlation. *Arnaud Le Troter, Shahram Attarian, Jean Pouget, David Bendahan, Emmanuelle Salort-Campana, Emilie Lareau-trudel (Marseille, France)*

PS3-294/#54 Sporadic late onset nemaline myopathy with MGUS: long term follow-up after SCT. *Olivier Benveniste, Monique Minnema, Henk Lokhorst, Martin Lammens, Wouter Meersseman, Michel Delforge, Thierry Kuntzer, Jan Novy, Thomas Pabst, Françoise Bouhour, Norma Romero, Véronique Leblond, Peter Van den Bergh, Baziel Van Engelen, Bruno Eymard, Nicol Voermans (Nijmegen, The Netherlands)*

PS3-286/#194 Clinical patterns in NT5C1A antibody positive sporadic Inclusion Body Myositis patients compared to seronegative patients. *Usman Alam, Tiyonnoh Cash, Farzin Pedouim, Sameen Enam, Farah Mozaffar, Tahseen Mozaffar, Namita Goyal (USA)*

PS3-272/#246 MDA-5 associated myositis : towards a molecular and morphological definition of a distinct entity. *Gaelle Leroux, Aude Rigolet, Baptiste Hervier, Miguel Hie, Nicolas Limal, Peter Hufnagl, Norman Zerbe, Thierry Maisonobe, Alain Meyer, Yurdagul Uzunhan, Francois-Jérome Authier, Jessie Aouizerate, Serge Herson, Olivier Benveniste, Werner Stenzel, Yves Allenbach (Berlin, Germany)*

PS3-291/#505 HMGB1 and RAGE expression in skeletal muscle inflammation: implications for protein accumulation in inclusion body myositis (IBM). *Ingrid E. Muth, Konstanze Kleinschnitz, Peter Balcarek, Arne Wrede, Stephan Zierz, Reinhard E. Voll, Marinos C. Dalakas, Jens Schmidt (Göttingen, Germany)*

PS3-277/#300 Clinical presentation and response to treatment of patients with the necrotizing immune-mediated myopathy associated with statins. *Josep M. Grau, José C. Milisenda, Albert Selva-O'Callaghan, Ricardo A. Losno, Alba Jerez, Marc Catalán, Pedro J. Moreno (Barcelona, Spain)*

PS3-311/#381 Seronegative myasthenia gravis- clinical and serological features. *Inga Koneczny, Leslie Jacobsen, David Beeson, Angela Vincent, Saif Huda (Oxford, UK)*

PS3-307/#272 Late-onset and very late-onset non-thymomatous anti-acetylcholine receptor antibody positive generalized myasthenia gravis: Clinical features. *Natalia A Juliá, Maria A Albertí, Inmaculada Pagola, Monica Povedano, Jordi Montero, Juan A Martínez-Matos, Antonio Martínez-Yélamos, Carlos Casasnovas, Christian Homedesc (Llobregat, Spain)*

PS3-313/#423 Prognostic factors in autoimmune myasthenia gravis.
Robert De Meel, Sander Lipka, Erik Van Zwet, Erik Niks, Jan Verschuuren (Leiden, The Netherlands)

PS3-310/#374 Epitope spreading is rare in MuSK myasthenia gravis. *Anna-Fleur Vink, Ricardo Rojas Garcia, Jordi Diaz Manera, Rinse Klooster, Kirsten Straasheijm, Erik Niks, Isabel Illa, Silvère van der Maarel, Jan Verschuuren, Maartje Huijbers (Leiden, The Netherlands)*

PS3-314/#443 HLA-DRB1*01 in late onset Myasthenia gravis. *Dina Lopes, Ana Martins Silva, Andreia Bettencourt, Isabel Moreira, Sandra Bras, Barbara Leal, Paulo Pinho Costa, Berta Martins Silva, Maria Isab Leite, Ernestina Santos (Porto, Portugal)*

PS3-320/#109 Missense mutations of agrin are responsible for a presynaptic form of congenital myasthenic syndrome with distal myopathy. *Amina Chaouch, Torberg Torbergsen, Stéphanie Godard-Bauché, Elodie de Bruyckere, Marie-José Fontenille, Morten Horn, Marijke van Ghelue, Yasmin Issop, Daniel Cox, Juliane S Müller, Christine Ioos, Annie Barois, Guy Brochier, Emmanuel Fournier, Daniel Hantaï, Angela Abicht, Marina Dusl, Steve H Laval, Helen Griffin, Bruno Eymard, Hanns Lochmüller, Sophie Nicole (Paris, France)*

PS3-344/#426 Antibodies against paranodal proteins detect CIDP patients with specific clinical phenotypes and poor response to conventional therapies. *Rojas-Garcia Ricard, Nogales-Gadea Gisela, Diaz-Manera Jordi, Gallardo Eduard, Pardo Julio, Seró Laia, Ortega-Moreno Angel, Bárcena Jose Eulalio, Sedano Maria Jose, Berciano Jose, Blesa Rafael, Dalmau Josep, Illa Isabel, Querol Luis (Barcelona, Spain)*

PS4-431/#214 Skeletal muscle fatty degenerative changes can be evaluated both qualitatively and quantitatively from whole-body Dixon NMR images with an important gain in acquisition time. *Benjamin Marty, Pierre-Yves Baudin, Benjamin Robert, Alexey Shukelovich, Robert-Yves Carlier, Noura Azzabou, Pierre G Carlier (Paris, France)*

11:30 **POSTER SESSION 3** *(Authors on allocated panel - See Poster sessions list on page 29)*

VISIT OF THE EXHIBITION

12:30 **INDUSTRY SPONSORED SYMPOSIUM** (ROOM CLIO)

12:30 **INDUSTRY SPONSORED SYMPOSIUM** (ROOM CALLIOPE)

14:30 **THEMATIC WORKSHOPS - SESSION 5**

TW 5.1 PNS SESSION 1: PATHOGENESIS OF IMMUNE MEDIATED NEUROPATHIES (AUDITORIUM ATHENA)

Coordinator: Eduardo Nobile-Orazio (Milan, Italy)

5.1.1 **Immunology and the nerve.** *Hans-Peter Hartung (Dusseldorf, Germany)*
5.1.2 **Nodo-paranodopathy: beyond the classification of myelinopathy and axonopathy.** *Nobuhiro Yuki (Singapore, Singapore)*
5.1.3 **Conduction block: Schwann cell or Axon pathology?** *Satoshi Kuwabara (Chiba, Japan)*
5.1.4 **Nerve pathology: still helpful in the diagnosis of immune neuropathies?** *Jean-Michel Vallat (Limoges, France)*

TW 5.2 RECENT ADVANCES IN UNDERSTANDING ANTIGENIC TARGETS IN MYASTHENIA GRAVIS (AUDITORIUM HERMES)

Coordinator: Socrates Tzartos (Athens, Greece)

5.2.1 **Defects of immune regulation in Myasthenia Gravis patients with anti-acetylcholine receptor antibodies.** *Sonia Berrih-Aknin (Paris, France)*
5.2.2 **Effector mechanisms of autoantibodies in Myasthenia Gravis.** *Marc De Baets (Maastricht, The Netherlands)*
5.2.3 **New autoantigens in MG.** *Socrates Tzartos (Athens, Greece)*

TW 5.3 NEUROMUSCULAR DISORDERS OF MITOCHONDRIAL FUSION AND FISSION (ROOM CLIO)

Coordinator: Véronique Paquis-Flucklinger (Nice, France)

5.3.1 **Novel phenotypes associated with mitochondrial dynamics deficiency.** *Véronique Paquis-Flucklinger (Nice, France)*
5.3.2 **Neuromuscular complications in patients with autosomal dominant optic atrophy.** *Patrick Yu-Wai-Man (Newcastle, United Kingdom)*
5.3.3 **Mitochondrial division and mitophagy in the brain and heart.** *Hiromi Sesaki (Baltimore, USA)*

TW 5.4 METABOLIC NEUROPATHY (ROOM THALIE)

Coordinator: Eva L Feldman (Ann Arbor, USA)

5.4.1 **Underlying Metabolic Mechanisms in the Pathogenesis of Diabetic Neuropathy.** *Eva L Feldman (Ann Arbor, USA)*
5.4.2 **Metabolic neuropathy: Clinical manifestations and current treatment.** *Brian Callaghan (Ann Arbor, USA)*
5.4.3 **Advances in the diagnosis and clinical interventions for metabolic neuropathy.** *Gordon Smith (Salt Lake City, USA)*

TW 5.5 **NUCLEAR ENVELOPE / NUCLEAR MATRIX** (ROOM ERATO)

Coordinator: Howard Worman (New York, USA)

5.5.1 **Emerin-LAP1 Interaction and X-linked Emery-Dreifuss Muscular Dystrophy.** *Howard Worman (New York, USA)*
5.5.2 **Lamin A/C and striated muscle laminopathies.** *Antoine Muchir (Paris, France)*
5.5.3 **Abnormal nuclear mechanics and mechanotransduction in muscular laminopathies.** *Jan Lammerding (New York, USA)*

16:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

16:30 **THEMATIC WORKSHOPS - SESSION 6**

TW 6.1 **PNS SESSION 2: NEW OPTIONS IN THE TREATMENT OF IMMUNE MEDIATED NEUROPATHIES** (AUDITORIUM ATHENA)

Coordinator: David R Cornblath (Baltimore, USA)

6.1.1 **Guillain-Barré syndrome: how to assess and to treat individuals with poor prognosis.** *Peter A van Doorn (Rotterdam, The Netherlands)*
6.1.2 **When and how to treat CIDP?** *Eduardo Nobile-Orazio (Milan, Italy)*
6.1.3 **Multifocal motor neuropathy: IVIg and ...what else?** *David R Cornblath (Baltimore, USA)*
6.1.4 **Rituximab in anti-MAG neuropathies: Yes or No?** *Jean-Marc Léger (Paris, France)*

TW 6.2 **NEW THERAPIES IN MYASTHENIA GRAVIS** (AUDITORIUM HERMES)

Coordinator: Amelia Evoli (Rome, Italy)

6.2.1 **New therapies for myasthenia gravis: just for refractory disease.** *Amelia Evoli (Rome, Italy)*
6.2.2 **New issues in the rational of thymectomy.** *Rozen Le Panse (Paris, France)*
6.2.3 **Antigen-specific apheresis in myasthenia gravis.** *Socrates Tzartos (Athens, Greece)*

TW 6.3 **FOCUS ON NEW INSIGHTS IN METABOLIC MYOPATHIES** (ROOM CLIO)

Coordinator: Pascal Laforêt (Paris, France)

6.3.1 **New genes for muscle glycogenosis.** *Pascal Laforêt (Paris, France)*
6.3.2 **Contribution of exercise tests to study treatment and the phenotypes of metabolic myopathies.** *Nicolai Preisler (Copenhagen, Denmark)*
6.3.3 **Exercise-induced rhabdomyolysis: diagnostic guidelines and RYR1-related cases.** *Nicol Voermans (Nijmegen, The Netherlands)*

TW 6.4 **PATHOMECHANISMS UNDERLYING MOTOR NEURON DEATH AND NEW THERAPEUTIC APPROACHES IN ALS/NMD** (ROOM THALIE)

Coordinator: Pamela Shaw (Sheffield, United Kingdom)

6.4.1 **New therapeutic targets for neuroprotection in ALS/NMD.** *Pamela Shaw (Sheffield, United Kingdom)*
6.4.2 **Dysregulation of RNA processing in motor neuron degeneration.** *Michael Sendtner (Wuerzburg, Germany)*
6.4.3 **Cellular autonomy and motor neurone degeneration.** *Siddharthan Chandran (Edinburgh, United Kingdom)*

TW 6.5 **UBIQUITIN-PROTEASOME COMPLEX IN HEART AND SKELETAL MUSCLES** (ROOM ERATO)

Coordinator: Mathias Gautel (London, United Kingdom)

6.5.1 **Regulation of ubiquitin-mediated turnover of sarcomeric proteins.** *Mathias Gautel (London, United Kingdom)*
6.5.2 **UPS dysfunction in hereditary cardiomyopathy (MYBPC3 / LMNA).** *Lucie Carrier (Hamburg, Germany)*
6.5.3 **Ubiquitin Proteasome system and skeletal myopathies.** *Homa Tajshargi (Gothenburg, Sweden)*

07:00 **BREAKFAST SEMINAR 3** (ROOM CLIO)

Organization of the costameric structures in skeletal muscle. *Stéphane Vassilopoulos (Paris, France)*

08:00 **PLENARY LECTURE SESSION 4** (AUDITORIUM ATHENA)

THERAPY IN NEUROMUSCULAR DISEASES

Chairpersons: Eva L Feldman (Ann Arbor, USA), Arthur Burghes (Columbus, USA)

PL 4.1 **AAV mediated gene therapy in Duchenne Muscular Dystrophy : A translational approach.** *Laurent Servais (Paris, France)*
PL 4.2 **Antisense Oligonucleotide therapies for Duchenne Muscular Dystrophy.** *Francesco Muntoni (London, United Kingdom)*
PL 4.3 **RNA targeted therapies for Myotonic Dystrophy.** *Charles A Thornton (Rochester, NY, USA)*
PL 4.4 **Therapy for GNE myopathy (DMRV/hIBM).** *Ichizo Nishino (Tokyo, Japan)*

10:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

10:30 **POSTER FLASH SESSION 4** (AUDITORIUM ATHENA)

Moderators: Thomas Voit (Paris, France), Jean Pouget (Marseille, France)

PS1-87/#407 Pulmonary Function and Safety Results at Week 120 of Exon-Skipping Drug Eteplirsen from the Phase 2b Study in Patients with Duchenne Muscular Dystrophy (DMD). *J R Mendell, L P Lowes, L Alfano, J Saoud, Edward M Kaye (Columbus, USA)*

PS1-93/#495 Drisapersen treatment for Duchenne muscular dystrophy (DMD): results of a 96-week follow-up of an open-label extension study following two placebo-controlled trials. *Rosamund Wilson, Giles Campion, Thomas Voit (Paris, France)*

PS1-85/#404 A Novel First in Human Study Design to Establish Tolerability of a Target Dose to Treat Patients with Duchenne Muscular Dystrophy (DMD) Amenable to Exon 53 Skipping. *Francesco Muntoni, Eugenio Mercuri, Thomas Voit, Volker Straub, V Ricotti, M Pane, L Ferron, M Guglieri, Petra Duda, J Saoud, Edward M Kaye (London, UK)*

PS1-104/#113 Tricyclo-DNA: highly promising antisense oligonucleotides for exon-skipping approaches in Duchenne Muscular Dystrophy. *Graziella Griffith, Samir El-Andaloussi, Kariem Ezzat, Arran Babbs, Branislav Dugovic, Stefan Schuerch, Cyrille Vaillend, Kay Davies, Christian Leumann, Luis Garcia, Aurélie Goyenvalle (Montigny le bretonneux, France)*

PS4-413/#490 Intraspinal Stem Cell Transplantation in ALS: Results of a Phase 1/2 Clinical Trial. *Nicholas Boulis, Stephen Goutman, Karl Johe, Seward Rutkove, Parag Patil, Jonathan Glass, Eva Feldman (USA)*

PS4-391/#128 Neuroprotective effects of JGK-263 in transgenic SOD1-G93A mice of amyotrophic lateral sclerosis. *Yoon-Ho Hong, Da-Eun Jeong, Jee-Eun Kim, Ji-Sun Kim, Kee Hong Park, Mu-Seok Park, Je-Young Shin, Sung-Yeon Sohn, Jung-Joon Sung, Suk-Won Ahn (Seoul, Republic of Korea)*

PS4-424/#318 A Phase II study to assess safety and efficacy of olesoxime (TRO19622) in 3-25 year old Spinal Muscular Atrophy (SMA) patients. *Enrico Bertini, Bruno Scherrer, Rebecca Pruss, Patrick Berna, Valérie Cuvier, Wilfried Hauke, Eric Dessaud (Roma, Italy)*

PS2-222/#560 Preliminary clinical efficacy and safety of BMN 701, GILT-tagged recombinant human acid alpha glucosidase (rhGAA) in late onset Pompe disease: Results of an extension study. *Barry Byrne, Richard Barohn, Bruce Barshop, Drago Bratkovic, Tarekegn Hiwot, Derralynn Hughes, Pascal Laforet, Eugen Mengel, Mark Roberts, William Lang, Jonathan LeBowitz, Claude Desnuelle (Gainesville, USA)*

PS2-242/#313 Long-term neurologic and cardiac correction in the Pompe disease mice by intrathecal gene therapy. *Laurence Dubreil, Cynthia Robveille, Quentin Pascal, Johan Deniaud, Mireille Ledevin, Candice Babarit, Marion Fusellier, Yassine Mallem, Carine Ciron, Corinne Huchet, Catherine Caillaud, Marie-Anne Colle, Juliette Hordeaux (Nantes, France)*

PS2-257/#528 Co-administration of the pharmamcological chaperone AT2220 with recombinant human acid alpha-glucosidase as a potential next-generation enzyme replacement therapy for Pompe disease. *Franklin Johnson, John Flanagan, Lee Pellegrino, Rebecca Soska, Jessie Feng, Richard Lazauskas, Julie Yu, Richie Khanna, Russell Gotschall, Hung Do, Kenneth Valenzano, Su Xu (Cranbury, USA)*

PS2-233/#161 Anti-alglucosidase alfa antibodies and infusion-associated reactions in 73 treated adult Pompe patients. *Juna de Vries, Marianne Hoogeveen-Westerveld, Stephan Wens, Marian Kroos, Michelle Kruijshaar, Pieter van Doorn, Ans van der Ploeg, Pim Pijnappel, Esther Kuperus (Rotterdam, The Netherlands)*

PS2-260/#533 Early diagnosis and early treatment in LOPD: when asymptomatic patients should be treated. *Giancarlo LaMarca, Severo Pagliardini, Marco Spada, Cesare Danesino, Giacomo Comi, Elena Pegoraro, Giovanni Antonini, Gianni Marrosu, Rocco Liguori, Lucia Morandi, Maurizio Moggio, Roberto Massa, Sabrina Ravaglia, Antonino Di Muzio, Corrado Angelini, Massimiliano Filosto, Paola Tonin, Giuseppe Di Iorio, Serena Servidei, Gabriele Siciliano, Tiziana Mongini, Antonio Toscano, Olimpia Musumeci (Messina, Italy)*

PS2-172/#116 Chemical chaperone ameliorates pathological protein aggregation in plectin-deficient muscle. *Lilli Winter, Ilona Staszewska, Eva Mihailovska, Irmgard Fischer, Wolfgang H Goldmann, Rolf Schröder, Gerhard Wiche (Vienna, Austria)*

PS2-266/#234 A controlled Phase 2 study of extended release sialic acid (SA-ER) in GNE myopathy. *Yoseph Caraco, Lau Heather, Alan Pestronk, Perry Shieh, Alison Skrinar, Jill Mayhew, Julia Martinisi, Emil Kakkis, Zohar Argov (Jerusalem, Israel)*

PS2-248/#425 Deoxypyrimidine Monophosphates Treatment for Thymidine Kinase 2 Deficiency. *Beatriz Garcia-Diaz, Valentina Emmanuele, Luis Carlos Lopez Garcia, Saba Tadesse, Orhan H Akman, Kurenai Tanji, Catarina Quinzii, Michio Hirano, Caterina Garone (New-York, USA)*

PS3-278/#311 Effects of auto-antibodies anti- signal recognition particle (SRP) and anti-Hydroxyméthylglutaryl-CoA reductase (HMGCR) on muscle cells. *Olivier Benveniste, Gillian Butler-Browne, Louiza Arouche-Delaperche (Paris, France)*

PS3-292/#523 Molecular treatment effects of alemtuzumab in skeletal muscle from patients with IBM. *Konstanze Kleinschnitz, Goran Rakocevic, Marinos C. Dalakas, Jens Schmidt, Karsten Schmidt (Göttingen, Germany)*

PS3-329/#110 Anti-myelin-associated glycoprotein neuropathy - a carbohydrate polymer effectively blocks pathogenic anti-myelin-associated glycoprotein antibodies. *Fan Yang, Nicole Schaeren-Wiemers, Andreas J. Steck, Beat Ernst, Ruben Herrendorff (Basel, Switzerland)*

PS3-331/#184 Immunoglobulin treatment in patients with Multifocal Motor Neuropathy: Insights from the SIGNS Registry. *Martin Stangel, David Pittrow, Ulrich Baumann, Maria Fasshauer, Dörte Huscher, Marcel Reiser, Manfred Hensel, Michael Borte, Wilhelm Kirch, Ralf Gold, Claudia Sommer (Würzburg, Germany)*

11:30 **POSTER SESSION 4** *(Authors on allocated panel - See Poster sessions list on page 32)*

VISIT OF THE EXHIBITION

12:30 **INDUSTRY SPONSORED SYMPOSIUM** (ROOM CALLIOPE)

14:30 **THEMATIC WORKSHOPS - SESSION 7**

TW 7.1 CELL THERAPY IN NEUROMUSCULAR DISEASES (AUDITORIUM ATHENA)

Coordinator: Jennifer Morgan (London, United Kingdom)

7.1.1 **Mouse satellite cells.** *Jennifer Morgan (London, United Kingdom)*
7.1.2 **Cell therapy for muscular dystrophies: update on the autologous myoblast cell therapy trial for oculopharyngeal muscular dystrophy.** *Gillian Butler-Browne (Paris, France)*
7.1.3 **Human artificial chromosomes and iPS cells for ex vivo gene therapy of muscular dystrophies and beyond.** *Francesco Saverio Tedesco (London, United Kingdom)*

TW 7.2 NEW PATHOMECHANISMS OF AMYOTROPHIC LATERAL SCLEROSIS (ROOM CLIO)

Coordinator: Leonard H van den Berg (Utrecht, The Netherlands)

7.2.1 **Genetics of Amyotrophic Lateral Sclerosis.** *Leonard H van den Berg (Utrecht, The Netherlands)*
7.2.2 **Overlap between Amyotrophic Lateral Sclerosis and Frontotemporal Dementia.** *Orla Hardiman (Dublin, Ireland)*
7.2.3 **Molecular mechanisms in Amyotrophic Lateral Sclerosis.** *Kevin Talbot (Oxford, United Kingdom)*

TW 7.3 NEW THERAPIES IN METABOLIC MYOPATHIES (AUDITORIUM HERMES)

Coordinator: Antonio Toscano (Messina, Italy)

7.3.1 **Therapeutic approaches in Lipid Storage Myopathies (LSMs).** *Antonio Toscano (Messina, Italy),*
7.3.2 **Therapies in glycogen storage myopathies.** *Benedikt Schoser (Munich, Germany)*
7.3.3 **Experimental therapy in mitochondrial disorders.** *Massimo Zeviani (Cambridge, United Kingdom)*

TW 7.4 PARANEOPLASTIC PERIPHERAL NEUROPATHY (ROOM THALIE)

Coordinator: Bruno Giometto (Treviso, Italy)

7.4.1 **Paraneoplastic sensory neuropathies: clinical and pathophysiological aspects.** *Jean-Christophe Antoine (Saint-Etienne, France)*
7.4.2 **Update on Lambert-Eaton myasthenic syndrome.** *Jan Verschuuren (Leiden, The Netherlands)*
7.4.3 **Paraneoplastic dysautonomia (from the PNS-Euronetwork Database).** *Bruno Giometto (Treviso, Italy)*

TW 7.5 INTERNAL MUSCLE CELL STRUCTURE (ROOM ERATO)

Coordinator: Edgar Gomes (Paris, France)

7.5.1 **The molecular basis of the stereotyped muscle organization.** *Clara Franzini-Armstrong (Philadelphia, USA)*
7.5.2 **Molecular basis of sarcoplasmic reticulum organization in skeletal muscle fibers.** *Vincenzo Sorrentino (Siena, Italy)*
7.5.3 **Formation and maintenance of triads during development and pathological conditions.** *Edgar Gomes (Paris, France)*

16:00 **COFFEE BREAK - VISIT OF THE EXHIBITION AND POSTER ZONE**

16:30 **THEMATIC WORKSHOPS - SESSION 8**

TW 8.1 THERAPEUTIC APPROACH TARGETING THE HEART IN NEUROMUSCULAR DISEASES (ROOM CLIO)

Coordinator: Lucie Carrier (Hamburg, Germany)

8.1.1 **RNA-based or gene-based therapies.** *Lucie Carrier (Hamburg, Germany)*
8.1.2 **Prevention and treatment of heart involvement in muscle diseases.** *Denis Duboc (Paris, France)*
8.1.3 **Targeting HDACs (histone deacetylase) in cardiovascular disease.** *Joseph A Hill (Dallas, USA)*

TW 8.2 TREAT NMD ALLIANCE: OMICS TECHNOLOGIES FOR TRANSLATIONAL RESEARCH IN NEUROMUSCULAR DISORDERS (AUDITORIUM HERMES)

Coordinator: Hanns Lochmüller (Newcastle, United Kingdom)

8.2.1 **Genomics and gene discovery in hereditary neuropathies.** *Jan Senderek (Munich, Germany)*
8.2.2 **Proteomics and pathophysiological signatures in myofibrillar myopathies.** *Rudolf A Kley (Bochum, Germany)*
8.2.3 **Biomarker discovery in Duchenne Muscular Dystrophy.** *Annemieke Aartsma-Rus (Leiden, The Netherlands)*

TW 8.3 DIAGNOSTIC BOUNDARIES OF LIMB GIRDLE MUSCULAR DYSTROPHIES (AUDITORIUM ATHENA)

Coordinator: John Vissing (Copenhagen, Denmark)

8.3.1 **Proximal weakness and atrophy: is this a limb girdle muscular dystrophy?** *John Vissing (Copenhagen, Denmark)*
8.3.2 **Overlap of phenotypes in dominantly inherited limb girdle muscular dystrophies vs. other dystrophies.** *Corrado Angelini (Padua, Italy)*
8.3.3 **Phenotypic transitions from congenital to limb girdle muscular dystrophies.** *Francesco Muntoni (London, United Kingdom)*

TW 8.4 ARTHROGRYPOSIS (ROOM THALIE)

Coordinator: Anders Oldfors (Gothenburg, Sweden)

8.4.1 **Arthrogryposis a general overview and syndromes.** *Mar Tulinius (Gothenburg, Sweden)*
8.4.2 **Amyoplasia: current concepts.** *Eva Kimber (Uppsala, Sweden)*
8.4.3 **Distal arthrogryposis: a developmental myopathy.** *Anders Oldfors (Gothenburg, Sweden)*

18:30-19:00 **CLOSING CEREMONY** (AUDITORIUM ATHENA)

POSTERS

POSTER SESSION1

PS1-1 / #138 - Role of Serum response factor in muscular satellite cells.
Voahangy Randrianarison-Huetz, Aikaterini Papaefthymiou, Laura Collard, Ulduz Faradova, Athanassia Sotiropoulos

PS1-23 / #464 - Novel gene networks and biological pathways regulated by FoxO in skeletal muscle during cancer cachexia. *Sarah Judge, Brandon Roberts, Chia-Ling Wu, Adam Beharry, Leonardo Ferreira, Susan Kandarian, Andrew Judge*

PS1-13 / #379 - Cytoskeleton network regulation and nuclear positioning during muscle development. *Vincent Gache, Bruno Cadot, Edgar Gomes*

PS1-27 / #406 - Characterization of the trafficking and functional properties of the muscle specific long STIM1 isoform.
Sophie Sauc, Laurent Bernheim, Pierre Cosson, Nicolas Demaurex, Maud Frieden

PS1-5 / #487 - Muscle Ring Finger-1 (MuRF1) activity is present in cardiac mitochondria and regulates reactive oxygen species production in vivo. *Taylor Mattox, Martin Young, Mathias Gautel, Ethan Anderson, Monte Willis*

PS1-25 / #258 - Defective mechanosensing responses are associated with altered Yes-associated protein (YAP) signaling in myoblasts from human muscular dystrophies resulting from mutations in LINC-complex associated proteins. *Martina Fischer, Tsolere Arakelian, Hélène Duchemin, Anne T Bertrand, Kamel Mamchaoui, Anne Bigot, Simindokht Ziaei, Thomas Voit, Gisèle Bonne, Catherine Coirault*

PS1-24 / #486 - Mrtf/Srf are mediators of mechanotransduction in skeletal muscle.
Alessandra Pincini, Laura Collard, Lorraine Montel, Sylvie Hénon, Athanassia Sotiropoulos

PS1-126 / #356 - Full-Length Dysferlin Expression Driven by Engineered Human Dystrophic Blood-Derived CD133+ Stem Cells. *Mirella Meregalli, Clementina Sitzia, Claire Navarro, Andrea Farini, Erica Montani, Nicolas Wein, Paola Razini, Cyriaque Beley, Letizia Cassinelli, Marzia Belicchi, Dario Parazzoli, Luis Garcia, Yvan Torrente*

PS1-20 / #107 - The Effect Of Aerobic Training And Detraining On Mitochondrial Function In Aging. *Søren Pete Andersen, Frank Thøgersen, Marie Loui Sveen, Mette Cath Ørngreen, Thomas Krag, John Vissing, Tina Dysga Jeppesen*

PS1-37 / #84 - The Wnt receptor Frizzled 3 is required for nerve-muscle target recognition process during neuromuscular junction formation.
Julien Messéant, Perrine Delers, Claire Legay, Laure Strochlic

PS1-42 / #422 - Neuromuscular junction integrity is dependent on rapsyn/AChR complex-intermediate filament linkage via plectin.
Eva Mihailovska, Marianne Raith, Ruth Herbst, Gerhard Wiche

PS1-99 / #551 - Comparison of rAAV6 and rAAV9 transduction in canines via jugular vein infusion. *Jeffrey Chamberlain, Jane Seto, Julian Ramos, Stephen Hauschka, Guy Odom*

PS1-39 / #215 - Integrins are required for synaptic transmission and development of the neuromuscular junction. *Jacob Ross, Richard Webster, Tanguy Lechertier, Francesco Muntoni, Jennifer Morgan, Kairbaan Hodivala-Dilke, Francesco Conti*

PS1-41 / #326 - The Molecular Machinery that Orchestrates Neuromuscular Junction Remodeling. *Tomasz Prószyski, Pawe Niewiadomski, Joshua Sanes*

PS1-87 / #407 - Pulmonary Function and Safety Results at Week 120 of Exon-Skipping Drug Eteplirsen from the Phase 2b Study in Patients with Duchenne Muscular Dystrophy (DMD).
J R Mendell, L P Lowes, L Alfano, J Saoud, Edward M Kaye

PS1-93 / #495 - Drisapersen treatment for Duchenne muscular dystrophy (DMD): results of a 96-week follow-up of an open-label extension study following two placebo-controlled trials. *Thomas Voit, Rosamund Wilson, Giles Campion*

PS1-85 / #404 - A Novel First in Human Study Design to Establish Tolerability of a Target Dose to Treat Patients with Duchenne Muscular Dystrophy (DMD) Amenable to Exon 53 Skipping. *Francesco Muntoni, Eugenio Mercuri, Thomas Voit, Volker Straub, V Ricotti, M Pane, L Ferron, M Guglieri, Edward M Kaye, Petra Duda, J Saoud*

PS1-104 / #113 - Tricyclo-DNA: highly promising antisense oligonucleotides for exon-skipping approaches in Duchenne Muscular Dystrophy. *Aurélie Goyenvalle, Graziella Griffith, Samir El-Andaloussi, Kariem Ezzat, Arran Babbs, Branislav Dugovic, Stefan Schuerch, Cyrille Vaillend, Kay Davies, Christian Leumann, Luis Garcia*

PS1-10 / #115 - Key role of EGFR in human primary myoblasts differentiation.
Laurent Bernheim, Stephane Konig, Julie Perroud

PS1-100 / #55 - Autosomal Recessive Limb Girdle Muscular Dystrophy: Prospective study and characterization of 280 cases by Immunohistochemistry and Immunoblotting.
Atchayaram Nalini, Gayathri Narayanappa, Bharath MM Srinivas, Sunitha Balaraju, Polavarapu Kiran, Modi Sailesh

PS1-101 / #77 - LAMA2-related muscular dystrophy- frequency and phenotype.
Nicoline Løkken, A. Peter Born, John Vissing

PS1-102 / #83 - Beta-sarcoglycanopathy: any longer an "orphan" disease? The Family Group of Beta-sarcoglycanopathy NPO was born in 2013.
Beatrice Vola, Roberto Maggi, Massimiliano Cerletti, Paola Bonetti

PS1-103 / #105 - Limb-girdle muscular dystrophies in the Czech Republic. Lenka Fajkusová, *Kristyna Stehlíková, Daniela Skálová, Lenka Mrázová, Petr Vondráek, Radim Mazanec, Stanislav Voháka, Markéta Hermanová, Josef Zámeník, Jana Haberlová*

PS1-105 / #119 - Executive function and visual memory computerized testing in myotonic dystrophies. *Stojan Peric, Elka Stefanova, Dusanka Savic-Pavicevic, Valerija Dobricic, Vesna Ralic, Jovan Pesovic, Ivana Novakovic, Vidosava Rakocevic-Stojanovic*

PS1-106 / #121 - Transcranial sonography in patients with myotonic dystrophy type 2.
Vidosava Rakocevic-Stojanovic, Stojan Peric, Dusanka Savic-Pavicevic, Jovan Pesovic, Aleksandra Pavlovic

PS1-107 / #122 - Assessment of pain in myotonic dystrophies. *Marina Peric, Stojan Peric, Valerija Dobricic, Dusanka Savic-Pavicevic, Vesna Ralic, Jovan Pesovic, Ivana Novakovic, Vidosava Rakocevic-Stojanovic*

PS1-108 / #135 - Oculopharyngeal Muscular Dystrophy in Population of the Czech Republic.
Radim Mazanec, Pavel Seeman, Eva Seemanova

PS1-109 / #158 - Cardiac arrhythmias in patients with laminopathies. *Michal Marchel, Agnieszka Serafin, Roman Steckiewicz, Agnieszka Madej-Pilarczyk, Krzysztof J Filipiak, Grzegorz Opolski*

PS1-11 / #211 - Control of transcription elongation is essential for cardiac and skeletal muscle development. *Yalda Jamshidi, Jaipreet Bharj, Merve Uysaloglu, Dongling Zheng, Jacob Ross, Francesco Muntoni, Daniel Osborn, Francesco Conti*

PS1-110 / #169 - Mutation in LMNA gene presenting as an overlapping syndrome with cardiomyopathy and muscle fibre type disproportion (FTD). *Lucia Ruggiero, Chiara Fiorillo, Alessandra Tessa, Fiore Manganelli, Rosa Iodice, Raffaele Dubbioso, Floriana Vitale, Filippo Maria Santorelli, Lucio Santoro*

PS1-111 / #171 - The mildest end of the dystroglycanopathy phenotypic spectrum: from asymptomatic hyperckaemia to limb girdle muscular dystrophy. *Chiara Fiorillo, Giacomo Brisca, Guja Astrea, Francesca Moro, Marina Pedemonte, Giorgia Negro, Carlo Minetti, Filippo Maria Santorelli, Claudio Bruno*

PS1-112 / #179 - Exploring mitochondrial dysfunction in CAPN3 related myopathy.
Chiara Fiorillo, Claudia Nesti, Mariachiara Meschini, Jacopo Baldacci, Stefano Doccini, Marina Mora, Claudio Bruno, Carlo Minetti, Filippo Maria Santorelli

PS1-113 / #210 - The relationship of calpain 3 and titin in the M-band. *Karine Charton, Jaakko Sarparanta, Anna Vihola, Laurence Suel, Nathalie Danièle, Peter Hackman, Bjarne Udd, Isabelle Richard*

PS1-114 / #212 - AAV-mediated transfer for ANO5-linked diseases.
Karine Charton, Laurence Suel, François Monjaret, Nathalie Bourg, Isabelle Richard

PS1-115 / #213 - Sarcoglycanopathy in Iran. *Yalda Nilipour, Shahriar Nafissi, Seyed Hass Tonekaboni, Parvaneh Karimzadeh, Reza Boostani, Gholamreza Zamani, Aryani Omid, Farah Ashrafzadeh, Ariana Kariminejad, Yousef Shafaghati, Masoud Hooshmand, Zahra Hadipour, Fatemeh Hadipour*

PS1-116 / #216 - Properties of mutant R388P of A-type lamins responsible for severe myopathy and lipodystrophy in patient skin fibroblasts and in HeLa cells. *Alice Barateau, Nolwenn Briand, Delphine Héron, Patrick Vicart, Corinne Vigouroux, Brigitte Buendia*

PS1-117 / #218 - Muscle pathology meets "next-gen sequencing data haystack": establishing the first diagnosis of DNAJB6-related myopathy in Austria.
Marcus Erdler, Reginald E Bittner, Wolfgang M Schmidt

PS1-118 / #221 - Determination of the best expression cassette for an AAV-mediated gene transfer – The case of a muscle specific protease, Calpain 3.
William Lostal, Carinne Roudaut, Laurence Suel, Isabelle Richard

PS1-119 / #222 - Hepatic AAV-mediated gene transfer to reduce immune responses against alpha-sarcoglycan. *Jérôme Poupiot, Isabelle Richard*

PS1-12 / #245 - Pericytes induce both satellite cell quiescence and differentiation during post-natal muscle growth.
Enis Kostallari, Yasmine Baba-Amer, Peggy Lafuste, Romain Gherardi

PS1-120 / #224 - Serum MYOM3 fragments as new biomarker for the follow-up of LGMD2D therapeutic treatment. *Jérôme Poupiot, Jérémy Rouillon, Bjarne Udd, Laurent Servais, Thomas Voit, Fedor Svinartchouk, Isabelle Richard*

PS1-121 / #225 - Limb Girdle Muscular Dystrophy 2I: Characterization of a new mouse model and testing AAV gene transfer.
Evelyne Gicquel, Karine Charton, Nathalie Daniele, Isabelle Richard

PS1-122 / #281 - Clinical characterization, measure of disability and mutational spectrum in a Chilean cohort of patients with dysferlinopathy.
Jorge A. Bevilacqua, Gabriella A. Di Capua, Lisanne Woudt, Martin Krahn, Claudia Castiglioni, Ricardo Hughes, Mario Campero, Alejandra Trangulao, Patricio González-Hormazábal, Raúl Godoy-Herrera, Nicolas Lévy, Andoni Urtizberea, Lilian Jara, Pablo Caviedes

PS1-123 / #304 - Molecular mechanisms and therapeutic approaches for sarcoglycanopathies. *Cécile Patissier, Jérôme Poupiot, Isabelle Richard*

PS1-124 / #320 - A simple questionnaire for screening patients with myotonic dystrophy type 1. *Tsuyoshi Matsumura, Takashi Kimura, Masayuki Nakamori, Katsuhisa Ogata, Harutoshi Fujimura, Masanori Takahashi P., Saburo Sakoda*

PS1-125 / #348 - New perspectives in LGMD-2D therapy: small molecules “to cure” the mutated alpha-sarcoglycan. *Elisa Bianchini, Romeo Betto, Dorianna Sandonà*

PS1-127 / #369 - A wide screening of the ANO5 gene by Next Generation Sequencing and Sanger sequencing confirms the clinical and genetic heterogeneity of LGMD2L and the incomplete penetrance. *Marco Savarese, Giuseppina Di Fruscio, Giorgio Tasca, Lucia Ruggiero, Sandra Janssens, Jan De Bleecker, Marc Delpech, Kathleen Claes, Olimpia Musumeci, Sabrina Sacconi, Lucio Santoro, Enzo Ricci, Luisa Politano, Vincenzo Nigro*

PS1-128 / #377 - Common and rare variants in genes related to limb girdle muscular dystrophies: questioning the disease-causing effect of previously reported genetic variants. Giuseppina Di Fruscio, Marco Savarese, Margherita Mutarelli, Vincenzo Nigro

PS1-129 / #413 - Natural history and peculiar aspects in LGMD2B. *Francesca Magri, Alessandra Govoni, Roberto Del bo, Maria Grazia D'angelo, Sandra Gandossini, Roberta Brusa, Irene Colombo, Isabella Moroni, Tiziana Mongini, Marina Mora, Corrado Angelini, Giuliano Tomelleri, Gabriele Siciliano, Antonio Toscano, Stefania Corti, Nereo Bresolin, Giacomo P. Comi*

PS1-130 / #446 - Emery-Dreifuss muscular dystrophy with LMNA mutation characterized by progressive cardiac conduction abnormality: a case report.
Katsuhisa Ogata, Mikiya Suzuki, Tomoyasu Hirano, Kana Yatabe, Toshiki Shigeyama, Kazunari Momma, Yuzo Tanaka, Yukiko Hayashi K, Ichizo Nishino, Ikuya Nonaka, Tadayuki Ishihara, Takuhisa Tamura, Mitsuru Kawai

PS1-131 / #484 - Caveolin 3 and lamin A/C: a common physiological way ?
Jean-Philippe Simon, Florian Barthelemy, Francesca Puppo, Sébastien Courrier, Marc Bartoli, Martin Krahn

PS1-132 / #498 - W2710X filamin C knock-in mice: a physiological model for filamin C-related myofibrillar myopathies. *Frédéric Chevessier-Tuennesen, Julia Schuld, Peter Van der Ven, Dieter Fürst, Rolf Schröder*

PS1-133 / #537 - A platform dedicated to the immortalization of human myoblasts isolated from patients with various neuromuscular disorders. *Anne Bigot, Kamel Mamchaoui, William Duddy, Elisa Negroni, Soraya Chaouch, Gillian Butler-Browne, Vincent Mouly*

PS1-134 / #550 - An International Web-based Registry for Dysferlinopathy Involving Participation of Patients and their Doctors. *Gaelle Blandin, Laura Rufibach, Brigitta von Rekowski, Céline Guien, Nicolas Lévy, Christophe Béroud, Martin Krahn*

PS1-14 / #394 - A modified cysteine knot ligand trap of the TGFB superfamily, ACE-083, increases muscle mass locally in mice. *Aaron Mulivor, Marishka Cannell, Monique Davies, Dianne Sako, Rita Steeves, Roselyne Castonguay, Samantha Wallner, Kathy Hevron, Danielle Bresnahan, Asya Grinberg, R. Scott Pearsall, Ravi Kumar*

PS1-15 / #398 - Dynamic of Triadin, a protein of the Calcium Release Complex.
Muriel Sebastien, Eric Denarier, Julie Brocard, Oriana Sarrault, Didier Grunwald, Isabelle Marty, Julien Faure

PS1-16 / #415 - Cytoskeleton composition of the human extraocular muscles.
Adrihan H. Janbaz, Eva Carlsson, Lena Carlsson, Mona Lindström, Fatima Pedrosa Domellöf

PS1-17 / #442 - Impairment of HACD1-dependent regulation of myoblast fusion leads to muscle hypotrophy. *Jordan Blondelle, Yusuke Ohno, Vincent Gache, Stéphane Guyot, Sébastien Storck, Nicolas Blanchard-Gutton, Marie Maurer, Laurent Guillaud, Geneviève Aubin-Houzelstein, Jean Demarquoy, Gemma Walmsley, Richard Piercy, Stéphane Blot, Akio Kihara, Laurent Tiret, Fanny Pilot-Storck*

PS1-18 / #473 - Changes in skeletal muscle structure and function following genetic inactivation of myostatin in rats. *Christopher Mendias, Jonathan Gumucio*

PS1-19 / #557 - A new phenotype-modifying mechanism that buffers nonsense mutation and avoids the extreme overgrowth of muscular tissue in the cattle.
Claire Bouyer, Ahmad Oulmouden

PS1-2 / #247 - Sdf-1 promotes BMSCs participation in regeneration of Pax7-/- mouse skeletal muscles. *Kamil Kowalski, Maria Ciemerych, Edyta Brzóska*

PS1-21 / #163 - New balances to control muscle mass and function: cross-talk of DHPR/ActIIBR in skeletal muscle pathophysiology. *Christel Gentil, Sestina Falcone, Gonzalo Jorquera, Mariana Casas, Helge Amthor, Thomas Voit, France Pietri-Rouxel*

PS1-22 / #305 - Protein biomarkers of Duchenne Muscular Dystrophy indicate a new component of the cellular pathogenesis : desintegration of the elastic mechanosensor network. *Jérémy Rouillon, Aleksandar Zocevic, Thibaut Léger, Camille GARCIA, Jean-Michel Camadro, Jérôme Poupiot, Isabelle Richard, Laurent Servais, Thomas Voit, Fedor Svinartchouk*

PS1-26 / #309 - The modulation of the NAD+ metabolism participates in the oxidative type fiber phenotype in skeletal muscle dysfunction in a mouse model of Emery-Dreifuss muscular dystrophy. *Nicolas Vignier, Maud Beuvin, Onnik Agbulut, Arnaud Ferry, Valerie Decostre, Alban Vignaud, Gisèle Bonne, Antoine Muchir*

PS1-28 / #507 - Depression of voltage-activated Ca2+ release in skeletal muscle by activation of a voltage-sensing phosphatase. *Christine Berthier, Candice Kutchukian, Clément Bouvard, Yasuchi Okamura, Vincent Jacquemond*

PS1-29 / #43 - Targeting miR-155 restores dysfunctional microglia and ameliorates disease in the SOD1 model of ALS. *Butovsky Oleg, Mark Jedrychowski, Ron Cialic, Muru Gopal, Pauline Wu, Camille Doykan, Zain Fanek, David Greco, Steven Gygi, James Berry, Merit Cudowicz, Howard Weiner*

PS1-3 / #303 - Control of the fate of human muscle stem cell by modulation of the in vitro microenvironment.
Claire Monge, Nicholas DiStasio, Anne Bigot, Vincent Mouly, Catherine Picart

PS1-30 / #183 - Decreased number of circulating NK cells and dramatic lack of INFgamma production in patients with antisynthetase syndrome. *Baptiste Hervier, Yves Allenbach, Fleur Cohen-Aubart, Werner Stenzel, Olivier Benveniste, Vincent Vieillard*

PS1-31 / #191 - Myasthenia Gravis associated with autoimmune Idiopathic Pulmonary Fibrosis. *Valeria Serban*

PS1-32 / #403 - Thymectomy and immune mechanisms in patients with myasthenia gravis. Cohort study in Czech Republic. *Michala Jakubíková, Michala Jakubíková, Jií Piha, Michaela Tyblová, Iveta Nováková, Helena Mare?ková, Jan Schutzner*

PS1-33 / #424 - Danger signals promoting innate immunity activation in Dermatomyositis. *Xavier Suárez-Calvet, Eduard Gallardo, Cinta Lleixà, Luis Querol, Ricard Rojas-Garcia, Jordi Díaz-Manera, Isabel Illa*

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PS1-34 / #290 - Age-related changes in central and peripheral nervous system associated with sarcopenia. *Vidya Nambiar, Vidya Nambiar, Tea Shavlakadze, Stuart Hodgetts, Alan Harvey, Miranda Grounds*

PS1-35 / #363 - Pre-symptomatic transcriptional profiling of differentially vulnerable motor neurons in the Smn2B/- mouse model of SMA.
Lyndsay Murray, Ariane Beauvais, Sabrina Gibeault, Rashmi Kothary

PS1-36 / #561 - Probing the Effect of Electrical Stimulation on the Formation of Neuromuscular Junctions in 2D and 3D. *Rodrigo Lozano, Brianna C. Thompson, Kerry J. Gilmore, Elise Stewart, Mario Romero-Ortega, Gordon G. Wallace*

PS1-38 / #156 - Wnt elicited molecular mechanisms during neuromuscular junction formation. *Julien Messeant, Perrine Delers, Carmen Marchiol, Gilles Renault, Claire Legay, Laure Strochlic*

PS1-4 / #344 - Changes in ALDH+ muscle cell populations with ageing in healthy and DMD patients and models. *Jessy Etienne, Cyril Catelain, Stéphanie Riveron, Stéphanie Lorain, Gillian Butler-Browne, Jean-Thomas Vilquin*

PS1-40 / #248 - MuSK-ColQ interaction and signalisation in synaptogenesis of the neuromuscular junction. *Alexandre Dobbertin, Claire Legay*

PS1-43 / #45 - Functional Motor Neuron Subtypes Generated from neurosphere drived adipose tissue for treatment of degenerative Motor Neuron diseases.
Marzieh Darvishi, Taki Tarihi

PS1-44 / #46 - In vitro induction of adipose derived stem cells into motoneurons characterized by calcium waves coupled/and with voltage dependent fluorescence.
Marzieh Darvishi, Taki Tarihi

PS1-45 / #112 - AAV9-mediated gene transfer of ?ARKct efficiently ameliorates cardiomyopathy in mice lacking dystrophin but not delta-sarcoglycan. *Ralf Bauer, Helene Enns, Andreas Jungmann, Barbara Leuchs, Christian Volz, Stefanie Schinkel, Philip Raake, Patrick Most, Hugo Katus, Oliver Müller*

PS1-46 / #142 - Mybpc3-targeted mice with hypertrophic cardiomyopathy show an impaired autophagic flux. *Sonia Singh, Saskia Schlossarek, Birgit Geertz, Lucie Carrier*

PS1-47 / #152 - Localization of Ankrd2 in PML bodies of skeletal muscle capillary cells. *Snezana Kojic, Sabine Krause, Ljiljana Rakicevic, Aleksandra Nestorovic, Jovana Jasnic Savovic, Johannes Vogel, Benedikt Schoser, Dragica Radojkovic, Maggie C Walter, Georgine Faulkner*

PS1-48 / #199 - Electromechanical delay components during skeletal muscle contraction and relaxation: novel physiological insights and possible application in Myotonic Dystrophies. *Fabio Esposito, Emiliano Cè, Susanna Rampichini, Eloisa Limonta, Barbara Fossati, Mauro Toffetti, Arsenio Veicsteinas, Giovanni Meola*

PS1-49 / #260 - Induction of neotendon formation and scleraxis expression in a supraphysiological model of tendon growth. *Jonathan Gumucio, Christopher Mendias*

PS1-50 / #358 - Comparisons between cellular candidates for mending cardiomyopathy in murine models. *Cyril Catelain, Stéphanie Riveron, Nathalie Mougenot, Michel Puceat, Gillian Butler-Browne, Gisèle Bonne, Jean-Thomas Vilquin*

PS1-51 / #419 - Fibromyalgia and hyperCKemia: electromyographic aspects, muscle biopsy and exercise related biochemical correlates. *Costanza Simoncini, margherita giorgetti, Laura Bazzichi, L Rossi, michelangelo mancuso, Giulia Ricci, Adele Servadio, erika schirinzi, Gabriele Siciliano*

PS1-52 / #64 - Effect of Sildenafil on skeletal and cardiac muscle function in Becker muscular dystrophy; a randomised, double blind, placebo-controlled crossover clinical trial. *Nanna Witting, Christina Kruuse, Bo Nyhuus, Kira Prahm, Gülsenay Citirak, Stine Lundgaard, Sebastian von Huth, Niels Vejlstrup, Thomas Krag, John Vissing*

PS1-53 / #75 - Post-exercise protein supplements improve muscle protein balance in Muscular Dystrophies. *Grete Andersen, Mette C Ørngreen, Nicolai R Preisler, Tina D Jeppesen, Simon Hauerslev, Thomas O Krag, Gerrit van Hall, John Vissing*

PS1-54 / #82 - Identification of components of the metabolic syndrome in patients with Duchenne/Becker muscular dystrophy. *Maricela Rodriguez-Cruz, Oriana Cruz-Guzman, Raúl Sánchez, Rosa Elena Escobar*

PS1-55 / #90 - Influence of a two-year steroid treatment on body composition as measured by Dual X-Ray Absorptiometry in boys with Duchenne Muscular Dystrophy. *Carole Vuillerot, Pierre Braillon, Stéphanie Fontaine-Carbonnel, Pascal Rippert, Elisabeth André, Jean Iwaz, Isabelle Poirot, Carole Bérard*

PS1-56 / #94 - Functional performance and muscle gene expression in dystrophic mdx mouse in relation to age and exercise: defects in mechanical-metabolic coupling.
Giulia Mar Camerino, Maria Cannone, Ada Maria Massari, Arcangela Giustino, Paola Mantuano, Roberta Fr Capogrosso, Anna Cozzoli, Annamaria De Luca

PS1-57 / #118 - Serum profiling identifies novel dystromiRs and cardiomyopathy-related miRNA biomarkers in GRMD dogs and DMD patients. *Laurence Jeanson-Leh, Julie Lameth, Soraya Krimi, Julien Buisset, Fatima Amor, Caroline Le Guiner, Inès Barthélémy, Laurent Servais, Stéphane Blot, Thomas Voit, David Israeli*

PS1-58 / #140 - Successful use of out-of-frame exon 2 skipping induces in vivo IRES-driven expression of a highly functional N-truncated dystrophin isoform: promising approach for treating other 5' dystrophin mutations. *Nicolas Wein, Adeline Vulin, Maria Sofi Falzarano, Cristina Al-Khalili Szigyarto, Baijayanta Maiti, Andrew Findlay, Kristin Heller, Mathias Uhlen, Baskar Bakthavachalu, Sonia Messina, Giuseppe Vita, Francesca Gualandi, Steve Wilton, Lin Yang, Diane Dunn, Daniel Schoenberg, Robert Weiss, Michael Howard, Alessandra Ferlini, Kevin Flanigan*

PS1-59 / #148 - Axial Muscle Involvment And Disease Progression In Dmd.
Jose Corderi, Alberto Dubrovsky, Lilia Mesa, Fernando Chloca, Agustin Jauregui, Patricia Marco, Maria Euge Gonzalez Toledo, Daniel Flores

PS1-6 / #488 - Non-targeted metabolomics analysis of Muscle Ring Finger-1 (MuRF1), MuRF2, and MuRF3 in vivo. *James Bain, Christopher Newgard, Ranjan Banerjee, Michael Muehlbauer, Monte Willis*

PS1-60 / #157 - Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. *Sabine Krause, Nikolai Klymiuk, Andreas Blutke, Alexander Graf, Katinka Burkhardt, Annegret Wuensch, Stefan Krebs, Barbara Kessler, Valeri Zakhartchenko, Mayuko Kurome, Elisabeth Kemter, Hiroshi Nagashima, Benedikt Schoser, Nadja Herbach, Helmut Blum, Rüdiger Wanke, Annemieke Aartsma-Rus, Eckhard Wolf, Maggie C. Walter, Hanns Lochmüller*

PS1-61 / #166 - Becker muscular dystrophy severity is linked to the structure of truncated dystrophins. *Aurélie Nicolas, Céline Raguénès-Nicol, Rabah Ben Yaou, Sarah Ameziane-Le Hir, Angélique Chéron, Véronique Vié, Mireille Claustres, France Leturcq, Olivier Delalande, Jean-François Hubert, Sylvie Tuffery-Giraud, Emmanuel Giudice, Elisabeth Le Rumeur*

PS1-62 / #182 - Sex effect on the efficacy of valproic acid therapy in mdx mice.
An-Bang Liu

PS1-63 / #187 - Muscular diseases diagnosis in West Africa: experience from four countries.
Maroufou Jules Alao

PS1-64 / #193 - Splice site strength and density of ESE and ESS motifs determine splicing pattern in the cases with splice site mutations in the dystrophin gene.
Tomoko Lee, Mariko Yagi, Yasuhiro Takeshima, Masafumi Matsuo, Kazumoto Iijima

PS1-65 / #196 - Expanded clinical spectrum of dystrophin copy number variants.
Katherine Howell, Robin Forbes, Trent Burgess, Desiree DuSart, Belinda Chong, Richard Leventer, Nigel Clarke, George McGillivray, Susan White, Steve Wilton, Catriona McLean, Monique Ryan

PS1-66 / #217 - Development of a novel approach using TALE nucleases to correct duplications in the dystrophin gene.
Sarah Farmer, Emma Wilson, Francesco Muntoni, Francesco Conti

PS1-67 / #227 - Morphologic and morphometric analysis of muscle degeneration in DMD: evolution in patients aged from 1 to 10 years. *Luisa Villa, Silvia Testolin, Lorenzo Peverelli, Patrizia Ciscato, Francesca Magri, Monica Sciacco, Giacomo Pi Comi, Maurizio Moggio*

PS1-68 / #240 - MiR-21 and miR-29 play opposing roles in the progression of fibrosis in Duchenne muscular dystrophy. *Simona Zanotti, Sara Gibertini, Paolo Savadori, Maurizio Curcio, Barbara Pasanisi, Lucia Morandi, Renato Mantegazza, Marina Mora*

PS1-69 / #275 - Loss of ambulation in the Cooperative International Neuromuscular Research Group (CINRG) Duchenne Muscular Dystrophy (DMD) cohort is synergistically influenced by glucocorticoid corticosteroid treatment and candidate genetic polymorphisms. Luca Bello, Akanchha Kesari, Heather Gordish-Dressman, Jaya Punetha, Erik Henricson, Tina Duong, Lauren Morgenroth, Elena Pegoraro, Avital Cnaan, Craig M. McDonald, Eric P. Hoffman

PS1-7 / #547 - Identify which population of muscle stem cells harbor the capacity to differentiate into autonomously beating pacemaker-like myocytes ?
Romain Davaze, Violeta Mitutsova, Mattia DiFrancesco, Pietro Mesirca, Matteo Mangoni, Ned J. Lamb, Anne Fernandez

PS1-70 / #279 - Muscular Dystrophy, antioxidants and metabolites: efficacy, mechanisms and optimal drugs. *Jessica Terrill, Hannah Radley-Crabb, Miranda Grounds, Peter Arthur*

PS1-71 / #285 - Forced exercise induces weakness in the mdx mouse. *Sivan Laban, Nurit Yanay, Moran Elbaz, Issa Butros, Malcolm Rabie, Yoram Nevo*

PS1-72 / #298 - Utrophin modulators to treat Duchenne muscular dystrophy (DMD): Future clinical trial plans for SMT C1100. *Francesco Muntoni, Stefan Spinty, Helen Roper, Imelda Hughes, Valeria Ricotti, Alison Bracchi, Graeme Horne, Jon Tinsley*

PS1-73 / #319 - Antisense oligonucleotide-mediated knockdown of TGF-?/myostatin type I receptor as a potential therapy for Duchenne and other muscular dystrophies.
Dwi U Kemaladewi, Svitlana Pasteuning, Sandra H van Heiningen, Johanna W van der Meulen, Gert-Jan van Ommen, Peter ten Dijke, Peter AC 't Hoen, Annemieke Aartsma-Rus, Willem M Hoogaars

PS1-74 / #333 - DMD/BMD patient registry in Japan: Remudy.
En Kimura, Harumasa Nakamura, Yukiko Hayashi K, Madoka Mori-Yoshimura, Reiko Shimizu, Hirofumi Komaki, Ichizo Nishino, Mitsuru Kawai, Shin'ichi Takeda

PS1-75 / #338 - New orally available compounds which modulate utrophin expression for the therapy of Duchenne muscular dystrophy (DMD).
Rebecca J. Fairclough, Sarah E. Squire, Noelia Araujo, Aini Vuorinen, Stephen G. Davies, Graham M. Wynne, Angela J. Russell, Kay E. Davies

PS1-76 / #354 - Assessing T cell-mediated immune response to dystrophin in the natural history of Duchenne muscular dystrophy. *Karen Anthony, Valeria Ricotti, Michela Guglieri, Laurent Servais, Thomas Voit, Katherine Bushby, Volker Straub, Jenny Morgan, Francesco Muntoni*

PS1-77 / #357 - Extracellular Adenosine-triphosphate (e-ATP) and purinergic signalling in inflammatory pathogenesis of dystrophin-deficient skeletal muscle. *Elisabetta Gazzerro, Simona Baldassari, Stefania Assereto, Chiara Panicucci, Chiara Fiorillo, Carlo Minetti, Elisabetta Traggiai, Fabio Grassi, Claudio Bruno*

PS1-78 / #359 - Restoration of mdx mice neuromuscular junctions as a mark of success of bone marrow stem cells therapy. *Anastasiia Sokolova, Natalia Timonina, Violetta Kravtsova, Igor Krivoi, Vyacheslav Mikhailov*

PS1-79 / #367 - Increased constitutive calcium entry via TRPC and TPV2 channels and decreased SERCA2a in mouse dystrophic cardiomyocytes.
Jose Javier Lopez, Elizabeth Aguettaz, Amal Houssaini, Regis Bobe, arnaud Ferry, Serge Adnot, Robert Hajjar, Larissa Lipskaia, Bruno Constantin

PS1-8 / #562 - Potential Link Between Alternative Splicing of Histidyl-tRNA Synthetase (HARS) and Inflammatory Myopathy. *Zhiwen Xu, Kyle P. Chiang, Feng Wang, Jie J. Zhou, Zhiyi Wei, Wing-Sze Lo, Xiang-Lei Yang, Leslie Nangle, Melissa Ashlock, Paul Schimmel, John Mendlein*

PS1-80 / #371 - SERCA2a is involved in the stabilization of plasma membrane calcium channels in human skeletal myotubes. *Amal Houssaini, Jose Javier Lopez, Regis Bobe, Arnaud Ferry, Serge Adnot, Robert Hajjar, Bruno Constantin, Larissa Lipskaia*

PS1-81 / #378 - Assessment of Upper Limb function in DMD patients:12 month changes.
Marika Pane, Elena Mazzone, Serena Sivo, Adele D'amico, Angela Berardinelli, Sonia Messina, Roberta Battini, Grazia D'angelo, Roberto De Sanctis, Lavinia Fanelli, Flaviana Bianco, Silvia Frosini, Elena Iotti, Giovanni Baranello, Patrizia Boffi, Lucia Morandi, Marina Pedemonte, Elena Pegoraro, Antonella Pini, Luisa Politano, Eugenio Mercuri

PS1-82 / #385 - Diapocynin, a putative NADPH oxidase inhibitor, ameliorates the phenotype of a mouse model of Duchenne muscular dystrophy.
Hesham Ismail Hamed, Leonardo Scapozza, Urs Ruegg, Olivier Dorchies

PS1-83 / #395 - Design of a confirmatory phase 3, multicenter, randomized, double-blind, placebo-controlled study of ataluren in patients with nonsense mutation Duchenne muscular dystrophy. *Allen Reha, Robert Spiegel, Gary Elfring, Jay Barth, Stuart Peltz*

PS1-84 / #401 - Preclinical Safety Profile Of Srp-4045, A Potential Phosphorodiamidate Morpholino Oligomer Treatment For Duchenne Muscular Dystrophy.
T Magee, JS Charleston, J Zhang, J Bhalli, H Kaur, J Walisser, P Sazani

PS1-86 / #405 - Phase 2b study of ataluren in nonsense mutation Duchenne muscular dystrophy: results across disease spectrum based on %-predicted 6MWD categories.
Craig McDonald, Erik Henricson, Richard T Abresch, Jay Barth, Allen Reha, Robert Spiegel, Stuart Peltz, Gary Elfring

PS1-88 / #409 - Phase 2b study of ataluren (PTC124®) in nonsense mutation Duchenne muscular dystrophy – results of a clinical efficacy robustness analysis.
Gary Elfring, Jay Barth, Allen Reha, Stuart Peltz, Craig McDonald

PS1-89 / #431 - Preclinical evaluation of tamoxifen and other selective estrogen receptor modulators in mdx5Cv dystrophic mice.
Olivier Dorchies, Julie Reutenauer-Patte, Sébastien Tardy, Hesham Ismail, Elyes Dahmane, Laurent Décosterd, Didier Picard, Urs Ruegg, Leonardo Scapozza

PS1-9 / #71 - Function of the H19 non coding RNA in muscle stem cells.
Clémence Martinet, Paul Monnier, Luisa Dandolo

PS1-90 / #471 - Effects of High Dose Gamma Irradiation on the EOM and Limb Muscles from a Mouse Model of Muscular Dystrophy. *Linda McLoon, Abby McDonald*

PS1-91 / #483 - Succesful pilot trial of L-arginine and metformin in Duchenne's muscular dystrophy. *Ulrike Bonati, Patricia Hafner, Beat Erne, Cornelia Neuhaus, Monika Gloor, Oliver Bieri, Erich Rutz, Stephan Frank, Arne Fischmann, Michael Sinnreich, Dirk Fischer*

PS1-92 / #485 - Comparison of Deflazacort and Prednisone in Duchenne Muscular Dystrophy. *Parvaneh Karimzadeh*

PS1-94 / #502 - Extracellular microRNAs: identity and function in relation to muscle regeneration and dystrophic pathology. *Anna Maria Lara Coenen-Stass, Thomas C. Roberts, Jennifer E Morgan, Yi Lee, Matthew J.A. Wood*

PS1-95 / #514 - Neuromuscular synaptic dysfunction in Duchenne muscular dystrophy mouse models.
E.M. van der Pijl, M. van Putten, J.J. Verschuuren, A.M. Aartsma-Rus, J.J. Plomp

PS1-96 / #525 - Duchenne muscular dystrophy (DMD): human IgG improves the performance of mdx mice in voluntary wheel running.
Jana Zschüntzsch, Florian Klinker, Yaxin Zhang, Gregor Makosch, Lars Klinge, Heinrich Brinkmeier, David Liebetanz, Jens Schmidt

PS1-97 / #527 - A Trial of Sildenafil treatment for patient with Duchenne muscular dystrophy. *Toshio Saito, Akie Kikuchi-Taura, Yumiko Iwata, Emi Muneshige, Hiroshi Yamamoto, Hiroaki Nishizono,, Tsuyoshi Matsumura, Harutoshi Fujimura, Saburo Sakoda*

PS1-98 / #538 - Development of Rimeporide, a NHE-1 inhibitor, for patients with Duchenne Muscular Dystrophy.
Myriam El Gaaloul, Florence Porte Thome, Béatrice Greco, Wolfgang Scholz, Caroline Kant

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POSTER SESSION 2

PS2-207 / #197 - MRI validation of a transcriptional cascade propagation model in FSHD muscular dystrophy. *BH Janssen, NBM Voet, AC Geurts, GW Padberg, BGM van Engelen, A Heerschap*

PS2-152 / #292 - Reducing dynamin 2 rescues myotubular myopathy in mice. *Belinda Cowling, Thierry Chevremont, Ivana Prokic, Christine Kretz, Arnaud Ferry, Catherine Coirault, Vincent Laugel, Norma Romero, Jocelyn Laporte*

PS2-171 / #106 - The pathogenesis of desminopathies: lessons from R350P desmin knock-in mice. *Christoph Clemen, Florian Stöckigt, Frederic Chevessier, Karl-Heinz Strucksberg, Lilli Winter, Harald Herrmann, Matthias Türk, Regine Schneider-Stock, Oliver Friedrich, Rainer Meyer, Oliver Müller, Jan Wilko Schrickel, Rolf Schröder*

PS2-136 / #192 - Impaired viability of muscle precursor cells in muscular dystrophy with glycosylation defects and amelioration of its severe phenotype by limited gene expression. *Motoi Kanagawa, Chih-Chieh Yu, Chiyomi Ito, So-ichiro Fukada, Tomoko Chiyo, Kazuhiro Kobayashi, Takashi Okada, Shin'ichi Takeda, Tatsushi Toda*

PS2-150 / #203 - Multimodal MRI and 31P-MRS investigations of the ACTA1(Asp286Gly) mouse model of nemaline myopathy provide evidence of impaired in vivo muscle function, altered muscle structure and disturbed energy metabolism. *Charlotte Gineste, Guillaume Duhamel, Yann Le Fur, Christophe Vilmen, Patrick J. Cozzone, David Bendahan, Julien Gondin*

PS2-180 / #451 - ZASP mutations in actin-binding domain cause disruption of skeletal muscle actin filaments in myofibrillar myopathy. *Xiaoyan Lin, Janelle Ruiz, Ilda Bajraktari, Rachel Ohman, Kenneth Fischbeck, Robert Griggs, Ami Mankodi*

PS2-204 / #497 - Digenic expression of sodium and chloride channel mutations in patients with non dystrophic myotonia. *Alain Furby, Savine Vicart, Jean-Philippe Camdessanche, Emmanuel Fournier, Stéphane Chabrier, Emmanuelle Lagrue, Renaud Touraine, Damien Sternberg, Bertrand Fontaine*

PS2-265 / #181 - Two founder mutations within GNE gene and high prevalence of GNE myopathy identified in North of Britain. *Oksana Pogoryelova, Amina Chaouch, Kathryn Brennan, Judith Hudson, Cheryl Longman, John McConville, Patrick Morrison, Maria Farrugia, Richard Petty, Willie Stewart, Fiona Norwood, Rita Horvath, Patrick Chinnery, Donald Costigan, John Winer, Tuomo Polvikoski, Estelle Healy, Anna Sarkozy, Michela Guglieri, Teresinha Evangelista, Michelle Eagle, Kate Bushby, Volker Straub, Hanns Lochmüller*

PS2-268 / #429 - Analysis of baseline sialic acid and NCAM data in GNE myopathy patients & mouse model. *Yiumo Chan, Paul Lee, Jaclyn Cadaoas, Gabrielle Morris, Emil Kakkis, Michel Vellard*

PS2-138 / #346 - Novel collagen VI chains in zebrafish skeletal muscle. *Laetitia Ramanoudjame, Claire Rocancourt, Jeanne Lainé, Arnaud Klein, Laura Lyphout, Corine Gartioux, Edor Kabashi, Xavier Cousin, Valérie Allamand*

PS2-226 / #85 - Pompe disease: pathophysiology and novel approaches to therapy. No authors listed for this abstract. *Nina Raben*

PS2-252 / #467 - Pathogenesis of the neuromuscular junction and motor neuron in Pompe disease. *Darin Falk, A. Gary Todd, Sooyeon Lee, David Fuller, Lucia Notterpek, Barry Byrne*

PS2-144 / #70 - Muscle histopathology in nebulin-related nemaline myopathy: ultrastrastructural findings correlated to disease severity and genotype. *Edoardo Malfatti, Vilma-Lott Lehtokari, Johann Böhm, Josine De Winter, Ursula Schaffer, Brigitte Estournet, Susana Quijano-Roy, Soledad Monges, Fabiana Lubieniecki, Rémi Bellance, Mai Thao Viou, Angeline Madelaine, Bin Wu, Analia Taratuto, Bruno Eymard, Katarina Pelin, Michel Fardeau, Coen Ottenheijm, Carina Wallgren-Pettersson, Jocelyn Laporte, Norma B Romero*

PS2-185 / #92 - Cardiac involvement in myotonic dystrophy: a nationwide cohort study. *Marie Lund, Lars Diaz, Matthis Ranthe, Helle Petri, Morten Duno, Inger Juncker, Hans Eiberg, John Vissing, Henning Bundgaard, Jan Wohlfahrt, Mads Melbye*

PS2-231 / #151 - A nationwide survey of Danon disease in Japan. *Kazuma Sugie, Hirofumi Komaki, Nobuyuki Eura, Ikuya Nonaka, Satoshi Ueno, Ichizo Nishino*

PS2-212 / #244 - Facioscapulohumeral muscular dystrophy type 1: quantitative MR imaging and clinical correlation. *Emilie Lareau-Trudel, Arnaud Le Troter, Shahram Attarian, Jean Pouget, David Bendahan, Emmanuelle Salort-Campana*

PS2-222 / #560 - Preliminary clinical efficacy and safety of BMN 701, GILT-tagged recombinant human acid alpha glucosidase (rhGAA) in late onset Pompe disease: Results of an extension study. *Barry Byrne, Richard Barohn, Bruce Barshop, Drago Bratkovic, Claude Desnuelle, Tarekegn Hiwot, Derralynn Hughes, Pascal Laforet, Eugen Mengel, Mark Roberts, William Lang, Jonathan LeBowitz*

PS2-242 / #313 - Long-term neurologic and cardiac correction in the Pompe disease mice by intrathecal gene therapy. *Juliette Hordeaux, Laurence Dubreil, Cynthia Robveille, Quentin Pascal, Johan Deniaud, Mireille Ledevin, Candice Babarit, Marion Fusellier, Yassine Mallem, Carine Ciron, Corinne Huchet, Catherine Caillaud, Marie-Anne Colle*

PS2-257 / #528 - Co-administration of the pharmamcological chaperone AT2220 with recombinant human acid alpha-glucosidase as a potential next-generation enzyme replacement therapy for Pompe disease. *Su Xu, Franklin Johnson, John Flanagan, Lee Pellegrino, Rebecca Soska, Jessie Feng, Richard Lazauskas, Julie Yu, Richie Khanna, Russell Gotschall, Hung Do, Kenneth Valenzano*

PS2-233 / #161 - Anti-alglucosidase alfa antibodies and infusion-associated reactions in 73 treated adult Pompe patients. *Juna de Vries, Esther Kuperus, Marianne Hoogeveen-Westerveld, Stephan Wens, Marian Kroos, Michelle Kruijshaar, Pieter van Doorn, Ans van der Ploeg, Pim Pijnappel*

PS2-260 / #533 - Early diagnosis and early treatment in LOPD: when asymptomatic patients should be treated. *Olimpia Musumeci, Giancarlo laMarca, Severo Pagliardini, Marco Spada, Cesare Danesino, Giacomo Comi, Elena Pegoraro, Giovanni Antonini, Gianni Marrosu, Rocco Liguori, Lucia Morandi, Maurizio Moggio, Roberto Massa, Sabrina Ravaglia, Antonino Di Muzio, Corrado Angelini, Massimiliano Filosto, Paola Tonin, Giuseppe Di Iorio, Serena Servidei, Gabriele Siciliano, Tiziana Mongini, Antonio Toscano*

PS2-172 / #116 - Chemical chaperone ameliorates pathological protein aggregation in plectin-deficient muscle. *Lilli Winter, Ilona Staszewska, Eva Mihailovska, Irmgard Fischer, Wolfgang H Goldmann, Rolf Schröder, Gerhard Wiche*

PS2-266 / #234 - A controlled Phase 2 study of extended release sialic acid (SA-ER) in GNE myopathy. *Zohar Argov, Yoseph Caraco, Lau Heather, Alan Pestronk, Perry Shieh, Alison Skrinar, Jill Mayhew, Julia Martinisi, Emil Kakkis*

PS2-248 / #425 - Deoxypyrimidine Monophosphates Treatment for Thymidine Kinase 2 Deficiency. *Caterina Garone, Beatriz Garcia-Diaz, Valentina Emmanuele, Luis Carlos Lopez Garcia, Saba Tadesse, Orhan H Akman, Kurenai Tanji, Catarina Quinzii, Michio Hirano*

PS2-135 / #117 - Satellite cells from Largemyd and Lama2dy2j/J: Are they different in their myogenic potential? *Paula Onofre-Oliveira, Poliana Martins, Amanda Lanzotti, Priscila Calyjur, Mariz Vainzof*

PS2-137 / #268 - Allele-specific silencing of a common dominant-negative COL6A3 mutation using siRNAs alleviates the phenotype of a cellular model of Ullrich congenital muscular dystrophy. *Véronique Bolduc, Yaqun Zou, Carsten G. Bönnemann*

PS2-139 / #360 - A novel mutation in COL6A1 caused recessive Bethlem myopathy. *Dmitry Vlodavets, Elena Belousova, Robert B. Weiss, Dmitry Kazakov*

PS2-140 / #509 - Body composition and energy expenditure in duchenne muscolar distrophy: longitudinal study. *Linda Berton, Silvia Sarti, Elena Ruggero, Elisa Frizzarin, luca bello, Andrea Barp, Elena Pegoraro, Giuseppe Sergi, Alessandra Coin*

PS2-141 / #549 - Inflammation in patients with Duchenne muscular dystrophy / Becker (DMD / BMD) with different nutritional status. *Oriana Cruz-Guzmán, Rosa Escobar, Maricela Rodriguez-Cruz, Raul Sanchez, Mariana Vega*

PS2-142 / #60 - Aerobic training in patients with congenital myopathy. *Gitte Hedermann, Christoffe Vissing, Nanna Witting, John Vissing*

PS2-143 / #65 - Endurance training in patients with collagen VI-related myopathy. *Gitte Hedermann, Christoffe Vissing, John Vissing*

PS2-145 / #99 - Adaptive role of mitochondrial changes in congenital central core myopathy. *Irina S. Vinogradskaya, Vladimir S Sukhorukov, Dmitry A. Kharlamov, Anatoly V. Brydun, Tatiana I. Baranich, Valeria V. Glinkina*

PS2-146 / #129 - Water-soluble fullerene improves skeletal muscle regeneration. *Akiko Ishii, Mizuko Yoshida, Norio Ohkoshi, Akira Tamaoka*

PS2-147 / #133 - Myosinopathies: Identification of MYH2, MYH3 and MYH8 cases by clinical, MR Imaging, histopathological and Electron microscopic studies.
Atchayaram Nalini, Gayathri Narayanappa

PS2-148 / #168 - Congenital titinopathies: an expanding spectrum.
Claire Chauveau, Carsten Bonnemann, Cedric Julien, Ay Lin Kho, Harold Marks, Beril Talim, Philippe Maury, Emmanuelle Uro-Coste, Alexander Alexandrovich, Anna Vihola, Livija Medne, Aileen Reghan Foley, Mariarita Santi, Bjarne Udd, Haluk Topaloglu, Steven A. Moore, Michael Gotthardt, Mark E. Samuels, Mathias Gautel, Ana Ferreiro

PS2-149 / #201 - Altered myosin cross-bridge cycling kinetics is associated with a paradoxical gain of muscle function in vivo in a mouse model of nemaline myopathy.
Charlotte Gineste, Coen Ottenheijm, Yann Le Fur, Sebastien Banzet, Emilie Pecchi, Christophe Vilmen, Patrick J. Cozzone, Nathalie Koulmann, Edna Hardeman, David Bendahan, Julien Gondin

PS2-151 / #223 - De novo mutation in RYR1. Case report. *Lenka Mrazova, Petr Vondracek, Lenka Fajkusova, Kristyna Stehlikova, Daniela Skalova, Marketa Hermanova, Hana Oslejskova*

PS2-153 / #299 - Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. *Johann Bohm, Valerie Biancalana, Edoardo Malfatti, Nicolas Dondaine, Catherine Koch, Nasim Vasli, Wolfram Kress, Matthias Strittmatter, Ana Lia Taratuto, Hernan Gonorazky, Pascal Laforêt, Thierry Maisonobe, Montse Olivé, Laura Gonzalez-Mera, Michel Fardeau, Nathalie Carrière, Pierre Clavelou, Bruno Eymard, Marc Bitoun, Joachim Weis, Jean-Louis Mandel, Norma Romero, Jocelyn Laporte*

PS2-154 / #331 - Selenoprotein N and oxidative stress regulate myogenesis and muscle stem cell differentiation. *Sandrine Arbogast, John Rowell, Alice Pannérec, Caroline Serreri, Charline Ramahefasolo, Giovanna Marazzi, David Sassoon, Ana Ferreiro*

PS2-155 / #351 - Amyoplasia : Muscle MRI findings and comparison with motor function. *Klaus Dieterich, Caroline Dubois, Frédérique Nugues, Adelaide Marquer, Marie Jaeger, Bernadette Berger, Marie-Christine Commare, Chantal Durand, Laurence Pittet-Barbier, Dominique Perennou, Pierre-Simon Jouk*

PS2-156 / #353 - Novel ECEL1 mutations and associated distal arthrogryposis phenotypes. *Brice Poreau, Klaus Dieterich, Nicole Monnier, Isabelle Marty, Pierre-Simon Jouk, Joël Lunardi, Julien Fauré*

PS2-157 / #370 - Inhibition of the ubiquitin proteasome system rescues defective SERCA1 protein causing Chianina cattle pseudomyotonia. *Roberta Sacchetto, Francesco Mascarello, Romeo Betto, Dorianna Sandonà, Elisa Bianchini*

PS2-158 / #388 - Successful cardiac transplantation in a young boy with congenital myopathy caused by ACTA 1 gene mutation. *Jana Haberlova, Richard Kirk, Hanns Lochmüller, Volker Straub, Kate Bushby*

PS2-159 / #399 - Identification by NGS of a novel MYH7 mutation in an Italian family affected by distal myopathy with multi-minicores. *Giacomo Brisca, Marco Savarese, Chiara Fiorillo, Paolo Broda, Giuseppina Di Fruscio, GianMichele Magnano, Carlo Minetti, Vincenzo Nigro, Claudio Bruno*

PS2-160 / #414 - Tubular aggregate myopathy is caused by mutations in the calcium sensor STIM1. *Johann Bohm*

PS2-161 / #416 - Molecular characterization of canine models for centronuclear myopathies. *Johann Bohm*

PS2-162 / #430 - Congenital myopathies and neonatal bone fractures. *Claudia Castiglioni, Alejandra Diaz, Verónica Ferrada, Alvaro Velásquez, Ricardo Erazo, Jorge Bevilacqua, Fabiana Fattori, Adele D`Amico, Enrico Bertini*

PS2-163 / #436 - Expanding the clinicopathological and genetic spectrum of RYR1 related Congenital Myopathies with cores and minicores: an Italian population study.
Elena Pegoraro, Denise Cassandrini, Guya Astrea, Valentina Codemo, Adele D'Amico, Chiara Fiorillo, Lorenzo Maggi, Francesca Magri, Marika Pane, Giorgio Tasca, Silvio Tosatto, Roberta Battini, Pia Bernasconi, Enrico Bertini, Giacomo P. Comi, Sonia Messina, Tiziana Mongini, Marina Mora, Lucia Morandi, Carlo Minetti, Enzo Ricci, Eugenio Mercuri, Filippo M. Santorelli, Claudio Bruno

PS2-164 / #450 - Congenital muscle disease with Rigid Spine, joint hyperlaxity and skin abnormalities: a novel phenotype and locus. *Laurianne Davignon, Claire Chauveau, Cedric Julien, Sylvie Odent, Leila Lazaro, Pascale Marcorelles, Jean-Paul Leroy, Ana Ferreiro*

PS2-165 / #454 - STIM1 mutations and muscle imaging in four Italian Tubular aggregate myopathy patients. *Giorgio Tasca, Adele D'Amico, Fabiana Fattori, Mauro Monforte, Enrzo Ricci, Enrico Bertini*

PS2-166 / #459 - Clinical heterogeneity in adult forms of FHL1 related myopathies. The "Institut de myologie" experience. *Rabah Ben Yaou, Tanya Stojkovic, Pascal Laforet, Alix De Becdelievre, Henri Marc Becane, Karim Wahbi, Carmen Navarro, Norma Beatriz Romero, Michel Fardeau, Pascale Richard, Denis Duboc, Gisele Bonne, Bruno Eymard, Rabah Ben Yaou*

PS2-167 / #76 - Induction of autophagy reduces aggregates in a cellular model of desminopathy. No authors listed for this abstract. *Gabriele Siciliano*

PS2-168 / #78 - Desmin, mechanics and myofibrillar myopathies. *Elisabeth Charrier, Atef Asnacios, Patrick Vicart, Sabrina Batonnet-Pichon, Sylvie Hénon*

PS2-169 / #79 - N-acetyl-L-cysteine prevents stress-induced desmin aggregation in cellular models of desminopathy.
Florence Delort, Bertrand-D Segard, Virginie Bailleux, Stéphanie Simon, Emilie Leccia, Blandine Gausseres, Fatma Briki, Patrick Vicart, Sabrina Batonnet-Pichon

PS2-170 / #104 - The German patient registry for protein aggregate myopathies.
Olivia Schreiber, Ralf Bauer, Oliver J. Mueller, Sabine Krause, Simone Thiele, Marcel Kiel, Rudolf Kley, Rolf Schroeder, Maggie C. Walter

PS2-173 / #220 - Differential proteomic analysis of abnormal intramyoplasmic aggregates in myotilinopathy. *Alexandra Maerkens, Montse Olivé, Sarah Feldkirchner, Joachim Schessl, Benedikt Schoser, Lev Goldfarb, Katrin Marcus, Matthias Vorgerd, Rudolf A. Kley*

PS2-174 / #259 - The "sphinx with dropped feet": a peculiar phenotype associated with heterozygous MYH7 mutation and core myopathy. *Ivana Dabaj, Edoardo Malfatti, Yann Pereon, Robert Carlier, Pascale Richard, Brigitte Estournet, Benjamin Dore, Marie Christine Durand, Nicole Monnier, Susana Quijano-Roy, Norma B. Romero*

PS2-175 / #327 - Distal myosin heavy chain-7 (thumb) myopathy due to the novel transition c.5566G>A with heterogeneous cardiac involvement.
Josef Finsterer, Claudia Stöllberger, Oliver Brandau, Franco Laccone

PS2-176 / #435 - A case of surplus protein myopathy: clinical and pathological characterization. *Giulia Ricci, Adele Servadio, Valentina Papa, Greta Alì, Costanza Simoncini, Lucia Chico, Giovanna Cenacchi, Gabriele Siciliano*

PS2-177 / #438 - Fhl1 W122S knock-in mice manifest late–onset mild myopathy.
Valentina Emmanuele, Akatsuki Kubota, Beatriz Garcia-Diaz, Caterina Garone, Hasan Orhan Akman, Kurenai Tanji, Catarina M Quinzii, Michio Hirano

PS2-178 / #444 - Recessive desmin deficiency myopathy with fatigability: Clinical features and response to salbutamol treatment. *Hacer Durmus*

PS2-179 / #448 - Characterization of ZASP-skeletal muscle actin interaction and its role in myofibrillar myopathy. *Janelle Ruiz, Xiaoyan Lin, Ilda Bajraktari, Robert Griggs, Rachel Ohman, Kenneth Fischbeck, Ami Mankodi*

PS2-181 / #489 - Distinct distal myopathy phenotype caused by common MATR3 gene mutation in a new family. *Kati Viitaniemi, Anni Evilä, Anna Vihola, Peter Hackman, Michio Hirano, Sanna Huovinen, Bashir Ayat, Kate Bushby, Bjarne Udd*

PS2-182 / #47 - Genetic Study of Brugada Syndrome in Tunisian population. *Kaabi Oldos, Ouali Sana, Abid Leila, Sahar Ben Kahla, Samir Kammoun, Rebai Tarek, Essia Boughzela, Bouayed Abdelmoula Nouha*

PS2-183 / #51 - Unusual manifestation in myotonic dystrophy type 1 mimicking bilateral internuclear ophthalmoplegia.
Chul-Hoo Kang, Sa-Yoon Kang, Ji Hoon Kang, Hong-Jun Kim, Jung Seok Lee, Sook Keun Song

PS2-184 / #563 - Understanding the pathogenic mechanisms underlying X-linked Charcot Marie Tooth neuropathy (CMTX6) caused by the R158H PDK3 mutation.
Gonzalo Perez-Siles, Eppie Yiu, David Chuang, Scott Tso, Aditi Kidambi, Adrienne Grant, Garth A. Nicholson, Marina L. Kennerson

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PS2-186 / #98 - The ultrasonographic evaluation of diaphragm in myotonic dystrophy. *Yoko Aburakawa, Takashi Kimura, Hideaki Kishi, Kenta Nomura, Kosuke Yoshida, Yasuhiro Suzuki, Kenji Kuroda, Osamu Yahara, Nao Kato, Chisato Murakami*

PS2-187 / #173 - Lower Urinary Tract and Bowel Dysfunction in Patients with Myotonic Dystrophy. *Stanislav Vohanka, Olesja Parmova, Jana Strenkova*

PS2-188 / #177 - MBNL1 autoregulates its function by binding to the 5'UTR of MBNL1 pre-mRNA and mRNA. *Patryk Konieczny, Katarzyna Taylor, ukasz Sznajder, Michal Kabza, Izabela Makalowska, Krzysztof Sobczak*

PS2-189 / #232 - Epidemiology of myotonic dystrophy in Bashkortostan (Russian Federation). *Regina Mukhametova, Elena Saifullina, Rim Magzhanov*

PS2-190 / #242 - Functional characterization of recessive ClC-1 mutations causing myotonia congenita. *Concetta Altamura, Simona Portaro, Norma Licata, Carmelo Rodolico, Olimpia Musumeci, Maria Maddalena Dinardo, Paola Imbrici, Antonio Toscano, Diana Conte Camerino, Jean-François Desaphy*

PS2-191 / #271 - Bacteraemic Pneumococcal Pneumonia. *Charles Feldman, Gregory Feldman*

PS2-192 / #330 - Predominantly myalgic phenotype caused by the p.A1156T mutation in SCN4A gene. *Johanna Palmio, Satu Sandell, Sini Penttilä, Bjarne Udd*

PS2-193 / #372 - Quantitative EMG analysis in myotonic dystrophy type 1 (DM1) and type 2 (DM2): comparative study. *Elzbieta Szmidt-Salkowska, Malgorzata Gawel, Anna Lusakowska, Monika Nojszewska, Anna Sulek, Wioletta Krysa, Marta Rajkiewicz, Andrzej Seroka, Anna Kaminska*

PS2-194 / #376 - Effects of acetazolamide on sarcolemma ionic conductances and excitability properties of skeletal muscle fibers as a possible therapeutic mechanism in disorders of skeletal muscle excitability. *Sabata Pierno, Maria Cannone, Kejla Musaraj, Jean-Francois Desaphy, Diana Conte Camerino*

PS2-195 / #383 - Functional and pharmacological characterization of a new hNav1.4 sodium channel mutation causing myotonia permanens. *Jean-Francois Desaphy, Roberta Carbonara, Anna Modoni, Adele D'Amico, Serena Pagliarani, Mauro Lo Monaco, Diana Conte Camerino*

PS2-196 / #387 - Clinical and molecular analysis of twenty five Algerian patients with myotonic dystrophy. *Karima Sifi, Nouredine Abadi, Abdelmadjid Hamri, Yamina Sifi, Michel Koenig*

PS2-197 / #408 - Direction of progression of motor impairment in DM1 patients; relation to CTG expansion. *Gro Solbakken, Tormod Hagen, Torunn Dahl Eikeland, Terje Nærland*

PS2-198 / #411 - DM1 and gender; how CTG expansion affect men and women differently. *Gro Solbakken, Tormod Hagen, Torunn Dahl Eikeland, Terje Nærland*

PS2-199 / #417 - Cis-elements regulating expression of muscleblind in Drosophila embryos. *Ariadna Bargiela, Estefania Herrero, Beatriz Llamusi, Ruben Artero*

PS2-200 / #421 - White matter changes and cognitive decline in DM1. *Sigrid Baldanzi, Leda Volpi, Paolo Cecchi, Serena Fabbri, Gianmichele Migaleddu, Anna Rocchi, Mirco Cosottini, Ilaria Pesaresi, Francesca Bevilacqua, Rita Lorio, Corrado Angelini, Gabriele Siciliano*

PS2-201 / #447 - Myotonic dystrophy type 2 coexisting with myasthenia gravis, pilomatricoma and lipoma in a family: a case report. *Anna Lusakowska, Malgorzata Szymczyk, Piotr Szczudlik, Anna Sulek, Marta Rajkiewicz, Wioletta Krysa, Anna Kaminska*

PS2-202 / #465 - Muscle channelopathies: clinical and genetic features in a large cohort of Italian patients. *Lorenzo Maggi, Raffaella Brugnoni, Lara Colleoni, Eleonora Canioni, Lucia Morandi, Renato Mantegazza, Pia Bernasconi*

PS2-203 / #470 - Higher risk of developing malignancies in myotonic dystrophy patients. *Roberto Fernandez-Torron, Miren Maneiro, Jose Ignacio Emparanza, Ana Maria Cobo, Juan Jose Poza Aldea, Juan Bautista Espinal, Miren Zulaika, Loreto Martorell, Adolfo Lopez de Munain*

PS2-205 / #524 - Cortical response during myotonia in myotonic dystrophy: an fMRI study. *Endre Pál, Arnold Tóth, Emese Lovadi, Ágnes Seb?k, Sámuel Komoly, József Janszky*

PS2-206 / #101 - A rare case of combined congenital pathology: facioscapulohumeral muscular dystrophy (FSHD) and cystic fibrosis. *Olga Klochkova, Alexey Kurenkov, Leila Namazova-Baranova, Ayaz Mamedyarov*

PS2-208 / #206 - Caveolar proteins: putative FSHD biomarkers? *Armelle Wauters, Alexandra Tassin, Baptiste Leroy, Steven Laval, Ruddy Wattiez, Alexandra Belayew*

PS2-209 / #207 - Development of fibrosis and its impact on muscle regeneration in FSHD muscle. *Céline Lancelot, Gilles Carnac, Paul Delrée, Denis Nonclercq, Frédérique Coppée, Alexandra Belayew*

PS2-210 / #208 - Study of atrophy in facioscapulohumeral muscular dystrophy. *Kelly Vancutsem, Sébastien Charron, Alexandra Belayew, Frédérique Coppée*

PS2-211 / #239 - Effects of vitamin C, vitamin E, zinc gluconate and selenomethionine supplementation on muscle function and oxidative stress biomarkers in patients with facioscapulohumeral dystrophy: a double-blind randomized controlled clinical trial. *Emilie Passerieux, Maurice Hayot, Gilles Carnac, Fares Gouzi, Fabien Pillard, Audrey Jaussent, Marie-Christine Picot, Koen Böcker, Gérald Hugon, Joel Pincemail, Jean O Defraigne, Theo Verrips, Jacques Mercier, Dalila Laoudj-Chenivesse*

PS2-213 / #249 - A mouse model of facioscapulohumeral muscular dystrophy. *Takako Jones, Chi Yan, Peter Jones*

PS2-214 / #250 - Regulation of DUX4 expression in facioscapulohumeral muscular dystrophy. *Charis Himeda, Céline Debarnot, Peter Jones, Takako Jones*

PS2-215 / #314 - The French National Registry for Facio-Scapulo-Humeral muscular Dystrophy: one year later. *Pauline Lahaut, Rafaelle Bernard, Katia Nehal, Gaëlle Blandin, Céline Guien, Karine Nguyen, Catherine Vovan, Karima Ghorab, Véronique Bombart, Pascal Cintas, Vincent Tiffreau, Marguerite Preudhomme, Arnaud Lacour, Marie-Christine Minot-Myhie, Tanya Stojkovic, Bruno Eymard, Emmanuelle Campana-Salort, Shahram Attarian, Françoise Bouhour, Claude Desnuelle, Christophe Beroud, Sabrina Sacconi*

PS2-216 / #316 - Toll-like receptors and innate immune system in the pathophysiology of oculopharyngeal muscular dystrophy. *Cristina Cappelletti, Franco Salerno, Eleonora Canioni, Lucia Morandi, Barbara Pasanisi, Lorenzo Maggi, Marina Mora, Dimos Kapetis, Barbara Galbardi, Renato Mantegazza, Pia Bernasconi*

PS2-217 / #332 - Back pain and paraspinal muscle involvement in patients with FSHD. *Julia Dahlqvist, Christoffer Vissing, Carsten Thomsen, John Vissing*

PS2-218 / #390 - Mitochondrial dysfunction reveals defective poly(A) tail regulation of specific mRNAs as a primary defect in oculopharyngeal muscular dystrophy. *Aymeric Chartier, Pierre Klein, Nicolas Barbezier, Teresa Gidaro, François Casas, Steven Carberry, Laurie Maynadier, George Dickson, Vincent Mouly, Gillian Butler-Browne, Kay Ohlendieck, Capucine Trollet, Martine Simonelig*

PS2-219 / #513 - Nuclear protein spreading: implication for pathophysiology of neuromuscular diseases. *Maxime Ferreboeuf, Virginie Mariot, Denis Furling, Gillian Butler-Browne, Vincent Mouly, Julie Dumonceaux*

PS2-220 / #515 - Gene therapy strategy for oculopharyngeal muscular dystrophy (OPMD). *Pierre Klein, Houria Bachtarzi, Alberto Malerba, Susan Jarmin, Sophie Perie, Jean Lacau St Guily, Mickael Graham, Gillian Butler-Browne, Vincent Mouly, George Dickson, Capucine Trollet*

PS2-221 / #516 - Cellular effectors of the exacerbated fibrosis in affected muscles of oculopharyngeal muscular dystrophy. *Elisa Negroni, Teresa Gidaro, Victorine Albert, Pierre Klein, Anne Bigot, William Duddy, Martine Oloko, Sophie Perie, Jean Lacau St Guily, Gillian Butler-Browne, Vincent Mouly, Capucine Trollet*

PS2-223 / #12 - Hypoparathyroidism as the First Manifestation of Kearns-Sayre Syndrome: A Case Report. Saghi Elmi

PS2-224 / #66 - Mitochondrial disease and risk of cancer. *Marie Lund, Mads Melbye, Lars Diaz, Morten Duno, Jan Wohlfahrt, John Vissing*

PS2-227 / #141 - X linked sideroblastic anemia and ataxia (XLSA/A) with mitochondrial myopathy and mental retardation caused by novel mutation of ATP-binding cassette transporter (ABCB7) gene. *Akihiro Hashiguchi, Yukie Inamori, Tadafumi Shiraishi, Itsuro Higuchi, Hiroshi Takashima*

PS2-228 / #144 - Severely impaired diaphragmatic function in Pompe disease visualized by cine-MRI. *Stephan Wens, Pierluigi Ciet, Adria Perez-Rovira, Karla Logie, Elizabeth Salamon, Piotr Wielopolski, Marleen de Bruijne, Michelle Kruijshaar, Harm Tiddens, Nadine van der Beek, Pieter van Doorn, Ans van der Ploeg*

PS2-229 / #147 - Enzime Replacement Therapy (Ert) In Pompe Disease – New Outcome Measures. *Alberto Dubrovsky, Roberto Peidro, Fernando Chloca, Agustin Jauregui, Graciela Brion, Marcelo Rugiero, Jose Corderi, Daniel Flores*

PS2-230 / #149 - Benefit of recombinant human acid alpha glucosidase treatment (Myozyme*) in late onset Pompe disease: about six cases with treatment for six months.
Fabien Zagnoli, Amelie Leblanc, Pascale Marcorelles

PS2-232 / #159 - Increased aortic stiffness and blood pressure in adults with Pompe disease. *Stephan Wens, Esther Kuperus, Francesco Mattace-Raso, Michelle Kruijshaar, Esther Brusse, Kees van Montfort, Marjan Scheltens-de Boer, Eric Sijbrands, Ans van der Ploeg, Pieter van Doorn*

PS2-234 / #164 - Histopathological, biochemical and clinical features in muscle biopsies of late-onset Pompe Disease patients before and after ERT. *Michela Ripolone, Raffaella Violano, Valeria Lucchini, Rubjona Xhani, Monica Sciacco, Dario Ronchi, Francesco Fortunato, Andreina Bordoni, Paola Tonin, massimilia Filosto, Stefano Previtali, Tiziana Mongini, Liliana Vercelli, Olimpia Musumeci, Corrado Angelini, Antonio Toscano, Costanza Lamperti, Marina Mora, Giacomo Pi Comi, Lucia Morandi, Maurizio Moggio*

PS2-235 / #170 - Mitochondria transfer from Wharton's jelly-derived stromal stem cell: a potential rescue strategy for mitochondrial diseases. *Tsu-Kung Lin, Hung-Yu Lin, Te-Yao Hsu, Chia-Wei Liou, Shang-Der Chen, Yao-Chung Chuang*

PS2-236 / #175 - A constitutive knock-out animal model for Glycogen storage disease type III. *Giacomo Comi, Serena Pagliarani, Gianna Ulzi, Sabrina Lucchiari, Fabrizio Seidita, Andreina Bordoni, Monica Nizzardo, Stefania Corti, Raffaella Violano, Michela Ripolone, Maurizio Moggio, Nereo Bresolin*

PS2-237 / #205 - Adult Polyglucosan Body Disease: clinical and histological heterogeneity of an Italian family. *Irene Colombo, Serena Pagliarani, Silvia Testolin, Ettore Salsano, Laura Napoli, Andreina Bordoni, Sabrina Salani, Elisabetta D'Adda, Lucia Morandi, Laura Farina, Maurizio Riva, Alessandro Prelle, Monica Sciacco, Giacomo Pi Comi, Maurizio Moggio*

PS2-238 / #276 - What factors are associated with the prevalence of sub-sarcolemmal mitochondrial aggregates (SSMA) in paediatric skeletal muscle? Examining the use and limitations of SSMA as a diagnostic muscle biopsy marker. *Andrea Cortese, Matt Ellis, Carl Fratter, Zoe Fox, Darren Chambers, Philip Hodsdon, Iain Hargreaves, Maria Kinali, Shamima Rahman, Caroline Sewry, Francesco Muntoni, Joanna Poulton, Rahul Phadke*

PS2-239 / #287 - Phosphoglucomutase type 1 (PGM1) deficiency bridges muscle glycogenosis and glycosylation disorders. *Thierry Dupre, Jean-Yves Hogrel, Tanya Stojkovic, Pascal Laforet, Isabelle Wargon, Catherine Sarret, Monique Piraud, François Petit*

PS2-240 / #294 - "Rolling in the deep": atypical presentations in late-onset pompe disease – beyond the "limb-girdle dystrophy phenotype". *Charles Lourenço, Vanessa Van der Linden, Claudia Sobreira, Wilson Marques Jr*

PS2-241 / #295 - Behr`s syndrome is a mitochondrial disease due to autosomal recessive mutations in the C12orf65 gene. *Rita Horvath, Angela Pyle, Venkateswaran Ramesh, Marina Bartsakoulia, Veronika Boczonadi, Agnes Heczegfalvi, Emma Blakely, Smertenko Tania, Jennifer Duff, David Moore, Patrick Yu Wai Man, Mauro Santibanez-Koref, Helen Griffin, Hanns Lochmüller*

PS2-243 / #322 - Assessing immune responses to recombinant human GAA (rhGAA) in late-onset Pompe disease (LOPD) patients. *Elisa Masat, Pascal Laforet, Damien Amelin, Kenza Laloui, Olivier Benveniste, Federico Mingozzi*

PS2-244 / #328 - Increased prevalence of malignancy in adult mitochondrial disorders. *Josef Finsterer, E. Krexner*

PS2-245 / #334 - A dominant mutation in CHCHD10 causes neurodegenerative disorder with mitochondrial DNA instability. *Samira Ait-El-Mkhadem, Sylvie Bannwarth, Annabelle Chaussenot, Konstantina Fragaki, Cécile Rouzier, Annie Verschueren, Jean Pouget, Véronique Paquis-Flucklinger*

PS2-246 / #352 - Exercise intolerance associated with ACAD9 mutations : a case report. *Annabelle Chaussenot, Konstantina Fragaki, Audrey Boutron, Christian Richelme, Sabrina Sacconi, Cécile Rouzier, Véronique Paquis-Flucklinger*

PS2-247 / #391 - Severe early onset cardiomyopathy in females with Danon disease not caused by skewed X-chromosome inactivation. *Carola Hedberg, Gyöngyvér Máthé, Kristjan Karason, Kate Thomson, Ingegerd Östman-Smith, Anders Oldfors*

PS2-249 / #437 - Acid -glucosidase rescue by splice-switching strategy using antisense oligonucleotides in patients with adult form of Pompe disease. *Aurelie Avril, Patrick Dreyfus, Branislav Dugovic, Pascal Laforêt, Christian Leumann, Catherine Caillaud, Luis Garcia*

PS2-250 / #441 - A multi-parametric protocol to study exercise intolerance in McArdle's disease. *Giulia Ricci, Federica Bertolucci, Costanza Simoncini, Ferdinando Franzoni, Riccardo Papi, Giovanni Pioggia, Annalisa LoGerfo, Gabriele Siciliano*

PS2-251 / #457 - A case of late-onset mitochondrial myopathy and ptosis due to a heterozygous DNA Polymerase gamma (POLG1) mutation.
Gudrun Zulehner, Marie-Therese Fischer, Martin Gencik, Gabor Kovacs, Jakob Rath, Uros Klickovic, Friedrich Zimprich, Hakan Cetin

PS2-253 / #469 - Clinical and genetic characteristics of chronic progressive external ophthalmoplegia (CPEO). *Jochen Schaefer, Daniela Leupold, Heinz Reichmann, Katrin Witte, Manja Weinhold, Sandra Jackson*

PS2-254 / #479 - Late onset NLSDM with novel mutations in the PNLPA2 gene in an Italian family. *Corrado Angelini, daniela Tavian, Sara Missaglia*

PS2-255 / #480 - Safety and efficacy of exercise training in 23 adults with Pompe disease receiving enzyme therapy. *Linda E.M. van den Berg, Marein M. Favejee, Stephan C Wens, Michelle E Kruijshaar, Stephan F.E. Praet, A.J. Reuser, Johannes B.J. Bussmann*

PS2-256 / #517 - The Influence of a Polymorphism in the Gene Encoding Angiotensin Converting Enzyme (ACE) on Treatment Outcomes in Late-Onset Pompe Patients Receiving Alglucosidase Alfa. *Rena Baek, Robert Pomponio, Rachel Palmer, Alison McVie-Wylie*

PS2-258 / #531 - Intrafamiliar phenotype variability related to a new mutation in SUCLA2 gene. *Juliana Gurgel-Giannetti, Beatriz Vilela, Caterina Garone, Ali Naini, Catarina Quinzi, Michio Hirano, Sergio Pena*

PS2-259 / #532 - Recurrent rhabdomyolysis due to muscle ?-enolase deficiency: expanding the clinical spectrum. *Olimpia Musumeci, Ros Quinlivan, Carmelo Rodolico, Stefen Brady, AnnaMaria Ciranni, Richard Kirk, Richard Godfrey, Elaine Murphy, Antonio Toscano*

PS2-261 / #539 - Acid phosphatase-positive rimmed vacuoles as useful marker in the diagnosis of adult-onset Pompe disease lacking specific clinical and pathological features. *Claire Dolfus, Francoise Chapon, Stephane Schaeffer, Jean Philippe Simon, Francois Leroy*

PS2-262 / #540 - Functional study of a germinal heterozygous 2bp-deletion in the SDHA gene identified in a patient with severe myopathy and late-onset cerebellar syndrome.
Frederique Savagner, Stephane Allouche, Jean Marc Constans, Danielle Herlicoviez, Francoise Chapon

PS2-263 / #543 - Molecular Basis of CoQ10 deficiency in the First Identified Patients.
Duygu Selcen, Andrew G. Engel

PS2-264 / #93 - X-linked myopathy with excessive autophagy (XMEA): NGS identifies a new in/del in the critic splicing region of the VMA21 gene. *Mariz Vainzof, Monize Lazar, Guilherme Yamamoto, Camila F. Almeida, Paula Onofre-Oliveira, Leticia Nogueira, Lydia Yamamoto U., Mayana Zatz, Rita Pavanello C.M., Helga C.A. Silva*

PS2-267 / #402 - Transthyretin Amyloidosis Outcomes Survey (THAOS): Early symptom presentation in hereditary transthyretin amyloidosis. *Teresa Coelho, Cecília Monteiro, Arnt Kristen, Merrill D. Benson, Onur N. Karayal, Rajiv Mundayat*

PS2-269 / #434 - Natural history study of GNE myopathy.
Nuria Carrillo-Carrasco, Lea Latham, Joseph Shrader, John Karl de Dios, Carla Ciccone, Frank Celeste, Chevalia Robinson, David Draper, Jahannaz Dastgir, Ami Mankodi, May Malicdan, Galen Joe, Marjan Huizing, John McKew, William Gahl

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PS3-323 / #519 - Molecular mechanisms of RAPSN mutations in congenital myasthenic syndromes. *David Beeson, Jonathan Cheung, Judith Cossins, Jacqueline Palace*

PS3-303 / #131 - In vitro characterization of satellite cells from myasthenic patients. *Mohamed Attia, Marie Maurer, Kamel Mamchaoui, Yoan Bismuth, Sylvain Bourgoin, Vincent Mouly, Gillian Butler-Browne, Sonia Berrih-Aknin*

PS3-285 / #165 - Phosphorylation of the autophagy receptor NBR1 by GSK3 modulates protein aggregation and is abnormal in muscles of sporadic inclusion body myositis patients. *Anne-Sophie Nicot, Francesca Lo Verso, Francesca Ratti, Fanny Pilot-Storck, Nathalie Streichenberger, Marco Sandri, Laurent Schaeffer, Evelyne Goillot*

PS3-273 / #261 - Redifining the immune histolochemical pattern of Anti-SRP auto-antibody positive patients: a subgroup present significant inflammation. *Yves Allenbach, Aude Rigolet, Tanya Stojkovic, Pascal Laforet, Antony Behin, Bruno Eymard, Norman Zerbe, Peter Hufnagl, Thierry Maisonobe, Kuberaka Mariampillai, Corinna Preusse, Serge Herson, Olivier Benveniste, Werner Stenzel*

PS3-274 / #264 - Myofiber HLA-DR expression is distinctive biomarker for antisynthetase myositis. *Jessie Aouizerate, Marie De Antonio, Thierry Maisonobe, Yasmine Baba-Amer, Romain K Gherardi, Francis Berenbaum, Loic Guillevin, Olivier Benveniste, Francois Jerome Authier*

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PS3-336 / #238 - Gene Expression Changes in Chronic Inflammatory Demyelinating Polyneuropathy Skin Biopsies. *Andreas Steck, Adrian Panaite, Stefania Puttini, Nicolas Mermod, Susanne Renaud, Thierry Kuntzer*

PS3-294 / #54 - Sporadic late onset nemaline myopathy with MGUS: long term follow-up after SCT. *Nicol Voermans, Olivier Benveniste, Monique Minnema, Henk Lokhorst, Martin Lammens, Wouter Meersseman, Michel Delforge, Thierry Kuntzer, Jan Novy, Thomas Pabst, Françoise Bouhour, Norma Romero, Véronique Leblond, Peter van den Bergh, Baziel van Engelen, Bruno Eymard*

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PS3-272 / #246 - MDA-5 associated myositis : towards a molecular and morphological definition of a distinct entity. *Yves Allenbach, Gaelle Leroux, Aude Rigolet, Baptiste Hervier, Miguel Hie, Nicolas Limal, Peter Hufnagl, Norman Zerbe, Thierry Maisonobe, Alain Meyer, Yurdagul Uzunhan, Francois-Jerome Authier, Jessie Aouizerate, Serge Herson, Olivier Benveniste, Werner Stenzel*

PS3-291 / #505 - HMGB1 and RAGE expression in skeletal muscle inflammation: implications for protein accumulation in inclusion body myositis (IBM). *Ingrid E. Muth, Konstanze Kleinschnitz, Peter Balcarek, Arne Wrede, Stephan Zierz, Reinhard E. Voll, Marinos C. Dalakas, Jens Schmidt*

PS3-277 / #300 - Clinical presentation and response to treatment of patients with the necrotizing immune-mediated myopathy associated with statins. *Pedro J. Moreno, Josep M. Grau, José C. Milisenda, Albert Selva-O'Callaghan, Ricardo A. Losno, Alba Jerez, Marc Catalán*

PS3-311 / #381 - Seronegative myasthenia gravis- clinical and serological features. *Saif Huda, Inga Koneczny, Leslie Jacobsen, David Beeson, Angela Vincent*

PS3-307 / #272 - Late-onset and very late-onset non-thymomatous anti-acetylcholine receptor antibody positive generalized myasthenia gravis: Clinical features. *Christian Homedes, Natalia A Juliá, Maria A Albertí, Inmaculada Pagola, Monica Povedano, Jordi Montero, Juan A Martínez-Matos, Antonio Martínez-Yélamos, Carlos Casasnovas*

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PS3-320 / #109 - Missense mutations of agrin are responsible for a presynaptic form of congenital myasthenic syndrome with distal myopathy. *Sophie Nicole, Amina Chaouch, Torberg Torbergsen, Stéphanie Godard-Bauché, Elodie De Bruyckere, Marie-José Fontenille, Morten Horn, Marijke Van Ghelue, Yasmin Issop, Daniel Cox, Juliane S Müller, Christine Ioos, Annie Barois, Guy Brochier, Emmanuel Fournier, Daniel Hantaï, Angela Abicht, Marina Dusl, Steve H Laval, Helen Griffin, Bruno Eymard, Hanns Lochmüller*

PS3-344 / #426 - Antibodies against paranodal proteins detect CIDP patients with specific clinical phenotypes and poor response to conventional therapies. *Querol Luis, Rojas-Garcia Ricard, Nogales-Gadea Gisela, Diaz-Manera Jordi, Gallardo Eduard, Pardo Julio, Seró Laia, Ortega-Moreno Angel, Bárcena Jose Eulalio, Sedano Maria Jose, Berciano Jose, Blesa Rafael, Dalmau Josep, Illa Isabel*

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PS3-300 / #72 - Autoimmunity - Myasthenia Gravis, Graves' disease and Vitiligo: a case report. *Valeria Serban*

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PS3-275 / #269 - Brain Perfusion Defects Correlate with Cognitive Deficits in Patients with Alum-induced Macrophagic Myofasciitis. *Emmanuel Itti, Mehdi Aoun-Sebaiti, Jessie Aouizerate, Nilusha Ragunathan-Thagarajah, Romain K Gherardi, Anne-Catherine Bachoud-Levi, Francois Jerome Authier*

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PS3-308 / #293 - Muscle cells undergo metabolic changes in Acetylcholine Receptor positive (AChR+) Myasthenia Gravis. *Marie Maurer, Sylvain Bougoin, Mohamed Attia, Jacky Bismuth, Rozen Le Panse, Gillian Butler-Browne, Sonia Berrih-Aknin*

PS3-288 / #302 - Mitochondrial DNA depletion in muscle from patients with sporadic inclusion body myositis. *Marc Catalán, Glòria Garrabou, Constanza Morén, Pedro Moreno, José Milisenda, Selva-O'Callahan Albert, Francesc Cardellach, Josep M. Grau*

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PS3-362 / #317 - Severe early onset Charcot-Marie-Tooth neuropathy caused by concomitant mutations in the MFN2 and GDAP1 revealed by Whole Exome Sequencing. *Anna Kostera-Pruszczyk, Joanna Kosinska, Agnieszka Pollak, Piotr Stawinski, Anna Walczak, Krystyna Wasilewska, Anna Potulska-Chromik, Piotr Szczudlik, Anna Kaminska, Rafal Ploski*

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PS3-363 / #345 - *Diagnostic neurosonography in demyelinating Charcot-Marie-Tooth Disease type 1A.*
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PS3-365 / #449 - *Autosomal dominant spinal muscular atrophy (ADSMA) with brisk tendon reflexes in a Czech patient with a de-novo complex mutation in the SETX gene. Petra Laššuthová, Dana Šafka Brožková, Jana Haberlová, Marcela Krtová, Pavel Seeman*

PS3-346 / #452 - *Clinical profile, treatment and outcomes of 121 Guillain-Barré syndrome episodes in 119 patients prospectively registered in the GBS National Czech Registry. Josef Bednarik, Tomáš Božovský, Edvard Ehler, Martin Forgá, Jana Haberlová, Jana Junkerová, Jilí Kuchylka, Radim Mazanec, Pavel Otruba, Martina Pátá, Petr Ridzo, Miroslav Škora, Jan Stank, Miloš Suchý, Peter Vaško, Alexander Vávra*

PS3-316 / #453 - *The Follow-up of the patients with Myasthenia Gravis in a period of 5 years. Altin Kuqo, Hariklia Doci, Liro Buda, Meri Papajani, Fjorda Myslymi, Serla Grabova, Aida Quka, Jera Kruja*

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PS3-347 / #463 - Unusual presentation of Axonal Neuropathy associated with MGUS IgM kappa paraproteins - Case Report. *Vanja Djuric V, Jelena Stamenovic, Stojanka Djuric S*

PS3-367 / #466 - Familial Amyloid Polineuropathy: efficacy of liver transplant versus tafamidis in nerve fiber function. *Marisa Brum, José Castro, Isabel Conceicao*

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PS3-369 / #493 - Neuropathy phenotype in Hereditary Transthyretin Amyloidosis.
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PS3-370 / #494 - Transthyretin cardiac amyloidosis (V122I) with clinical and histological evidence of amyloid neuropathy and myopathy. *Aisling Carr, Zane Jaunmuktane, David Hutt, S Brandner, Estelle Healy, Janice Holton, Julian Blake, Carol Whelan, A Wechalekar, Jullian Gilmore, Phillip Hawkins, Mary Reilly*

PS3-389 / #503 - Acquired idiopathic generalized anhidrosis – clinical, neurophysiological, pathological findings and treatment response on thirteen patients. *So-Hee Park, Sang-Beom Kim, Bum Chun Suh, Jeeyoung Oh*

PS3-348 / #504 - A case of Multifocal acquired demyelinating motor sensory neuropathy complicated by phrenic nerve palsy. *Sun Young Kim, Seok Jung Im*

PS3-349 / #511 - Primary neurolymphomatosis from low-grade B-cell lymphoma presenting as slowly progressive length dependent polyneuropathy.
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PS3-318 / #521 - Myasthenia gravis in senegalese children: 10 years follow up at Fann teaching hospital, Dakar. *Marieme Soda Diop, Moustapha Ndiaye, Anna Basse, Ndeye Fatou Ndoye, Lala Bouna Seck, Hawa Sidibe, Adjaratou Sow, Amadou Gallo Diop, Kamadore Toure, Mouhamadou Mansour Ndiaye*

PS3-283 / #529 - Case report of juvenile polymyositis with. *Pamela Rapiti, Anand Rapiti*

PS3-390 / #534 - Serial measurement of intraepidermal nerve fiber loss in critically ill patients. *Roman Kopacik, Josef Bednarik, Miroslav Skorna, Milena Kostalova, Eva Vlckova*

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PS3-324 / #544 - The spectrum of DOK7 congenital myasthenia in Northern Ireland. *Grace McMacken, Aisling Carr, Estelle Healy, Kiang Pang, Jacqueline Palace, David Beeson, John McConville*

PS3-298 / #548 - A case of rhabdomyolysis due to cyanide poisoning following the consumption of European black elderberries. *Uros Klickovic, Jakob Rath, Gudrun Zulehner, Hakan Cetin*

PS3-371 / #552 - Search for genetic modifiers of CMT1A and HNPP by evaluating the extremes of the clinical spectrum. *Barbara van Paassen, Fred van Ruissen, Camiel Verhamme, Karin van Spaendonck-Zwarts, Marianne de Visser, Frank Baas, Anneke van der Kooi*

PS3-351 / #553 - A Guillain-Barre syndrome epidemic following a Zika virus epidemic in French polynesia. *Frederic Ghawché, Philippe Larre, Stephane Lastere, isabelle leparc-goffart, Henri-Pier Mallet, Didier Musso, jean Neil, chantal sookhareea, Louise Watrin*

PS3-372 / #554 - Rasch-built overall disability scale for Charcot-Marie-Tooth disease (CMT-RODS). *Fleur Rövekamp, Barbara van Paassen, Wim Linssen, Michael E. Shy, Mary M. Reilly, Marianne de Visser, Ingemar Merkies, Anneke van der Kooi*

PS3-325 / #555 - Congenital myasthenic syndrome: identification of a MuSK mutation that results in exon 9 skipping and could impair the agrin system. *Jeanine Koenig, Emmanuelle Girard, Stéphanie Bauché, Pascale Richard, Valérie Risson, Thomas Simonet, Asma Ben Ammar, Guy Brochier, Frédéric Chevessier, Louis Viollet, Evelyne Goillot, Yuji Yamanashi, Laurent Schaeffer, Bruno Eymard, Daniel Hantaï*

PS3-326 / #556 - Congenital myasthenia due to AGRN c.5125G>C: does the mutated agrin perturb motoneuron differentiation? *Jeanine Koenig, Stéphanie Bauché, Emmanuelle Girard, Valérie Risson, Asma Ben Ammar, Pascale Richard, Evelyne Goillot, Antoine Taly, Laurent Schaeffer, Bruno Eymard, Daniel Hantaï*

PS3-319 / #559 - Myasthenia gravis with double antibodies positivity (AchRAb and anti-MuSK) associated with severe thymic maligrancy: description of 6 patients. *R. Ricciardi, M. Maestri, A. De Rosa, M. Lucchi, U. Bonuccelli, A. Mussi*

POSTER SESSION 4

PS4-393 / #146 - Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. *Kathrin Meyer, Laura Ferraiuolo, Carlos Miranda, Shibi Likhite, Arthur Burghes, Stephen Kolb, Brian Kaspar*

PS4-415 / #59 - The pathogenic contribution of astrocytic and muscular TWEAK to ALS pathology. *Melissa Bowerman, Céline Salsac, Emmanuelle Coque, Frédérique Scamps, Alexandre Brodovitch, Cedric Raoul*

PS4-455 / #308 - Targeted and exome sequencing for diagnosis and novel gene identification in congenital myopathies. *Valerie Biancalana, Johann Bohm, Osorio Lopes Abath Neto, Edoardo Malfatti, Nicolas Dondaine, Nasim Vasli, Norma Romero, Jocelyn Laporte*

PS4-420 / #172 - Muscle mitochondrial dysfunction in a large cohort of genetically-determined SMA patients. *Michela Ripolone, Dario Ronchi, Raffaella Violano, Dionis Vallejo, Emanuele Barca, Gigliola Fagiolari, Angela Berardinelli, Umberto Ballottin, Lucia Morandi, Marina Mora, Andreina Bordoni, Francesco Fortunato, Antonio Toscano, Monica Sciacco, Salvatore Di Mauro, Giacomo Pi Comi, Maurizio Moggio*

PS4-405 / #558 - ALS astrocytes kill motor neurons via ligation of death receptor 6 by a fragment of N-APP/APLP1. *Diane B. Re, Virginia Le Verche, Burcin Ikiz, Mariano Alvarez, Kristin Politi, Paschalis-Toma Doulias, Dimitra Papadimitriou, Todd Greco, Anatoly Nikolaiev, Andrea Califano, Harry Ischiropoulos, Manuel Than, Marc Tessier-Lavigne, Serge Przedborski*

PS4-439 / #361 - Upper limb muscle fat-water quantification MRI and clinical functional evaluation in non-ambulant Duchenne muscular dystrophy. *Valeria Ricotti, Matthew Evans, Christopher Sinclair, Jasper Morrow, Jordan Butler, Robert Janiczek, Michael Hanna, Paul Matthews, Tarek Yousry, Francesco Muntoni, John Thornton*

PS4-417 / #124 - Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. *Annie Laquérriere, Jérome Maluenda, Adrien Camus, Laura Fontenas, Klaus Dieterich, Flora Nolent, Jié Zhou, Nicole Monnier, Philippe Latour, Joel Lunardi, Monica Bayes, Pierre S Jouk, Damien Sternberg, Josiane Warszawski, Ivo Gut, Marie Gonzales, Marcel Tawk, Judith Melki*

PS4-431 / #214 - Skeletal muscle fatty degenerative changes can be evaluated both qualitatively and quantitatively from whole-body Dixon NMR images with an important gain in acquisition time. *Benjamin Marty, Pierre-Yves Baudin, Benjamin Robert, Alexey Shukelovich, Robert-Yves Carlier, Noura Azzabou, Pierre G Carlier*

PS4-413 / #490 - Intraspinal Stem Cell Transplantation in ALS: Results of a Phase 1/2 Clinical Trial. *Eva Feldman, Nicholas Boulis, Stephen Goutman, Karl Johe, Seward Rutkove, Parag Patil, Jonathan Glass*

PS4-391 / #128 - Neuroprotective effects of JGK-263 in transgenic SOD1-G93A mice of amyotrophic lateral sclerosis. *Yoon-Ho Hong, Da-Eun Jeong, Jee-Eun Kim, Ji-Sun Kim, Kee Hong Park, Mu-Seok Park, Je-Young Shin, Sung-Yeon Sohn, Jung-Joon Sung, Suk-Won Ahn*

PS4-424 / #318 - A Phase II study to assess safety and efficacy of olesoxime (TRO19622) in 3-25 year old Spinal Muscular Atrophy (SMA) patients. *Enrico Bertini, Eric Dessaud, Bruno Scherrer, Rebecca Pruss, Patrick Berna, Valérie Cuvier, Wilfried Hauke*

PS4-392 / #134 - The blood-spinal cord barrier is temporarily impaired in TDP-43 conditional knockout mice. *Shoichi Sasaki, Yohei Iguchi, Masahisa Katsuno, Gen Sobue*

PS4-394 / #174 - Satellite cell activation in muscles of terminal ALS patients. *Anton Tjust, Mona Lindström, Fatima Pedrosa Domellöf*

PS4-395 / #266 - human pluripotent stem cells : potential for neuro muscular diseases modeling? *Yves Maury, cécile Martinat, Stephane Nedelec*

PS4-396 / #297 - Adult neurogenesis in Amyotrophic Lateral Sclerosis patients with or without dementia. *Lucia Galan, Ulises Gomez-Pinedo, Antonio Guerrero-Sola, Alvaro Vela-Souto, Armando Martinez-Martinez, Maria Sol Benito-Martin, Alberto Rabano-Gutierrez, Jose Manuel Garcia-Verdugo, Jordi Matias-Guiu*

PS4-397 / #321 - Role of beta – Amyloid oligomeric (AbO) fraction in ALS disease. *Noelle Callizot, Philippe Poindron, Lucie Mottier*

PS4-398 / #339 - Diffusion magnetic resonance imaging: a promising novel tool for early detection and monitoring of motor neuron degeneration in ALS. *Stefania Marcuzzo, Victoria Moreno-Manzano, Ileana Zucca, Alessandro Scotti, Matteo Bigini, Silvia Bonanno, Barbara Galbardi, Dimos Kapetis, Pia Bernasconi, Renato Mantegazza*

PS4-399 / #382 - The Missing Factors Influencing Spinal And Bulbar Muscular Atrophy Phenotype: Evaluation Of Genetic Polymorphisms. *Cinzia Bertolin, Giorgia Querin, Maria Pennuto, Francesca Zoccarato, Elena Pegoraro, Cinzia Gellera, Gianni Sorarù*

PS4-400 / #386 - Human induced pluripotent stem cells as an in vitro model for investigating infantile-onset ascending hereditary spastic paralysis pathogenesis. *Sara D'Alessandro, Stefania Marcuzzo, Claudia Barzago, Silvia Bonanno, Dimos Kapetis, Barbara Galbardi, Giovanna Zorzi, Renato Mantegazza, Pia Bernasconi*

PS4-401 / #468 - Stem cell-derived motor neurons from spinal and bulbar muscular atrophy patients. *Christopher Grunseich, Kristen Zukosky, Ilona Kats, Laura Bott, Carlo Rinaldi, Kenneth Fischbeck*

PS4-402 / #475 - Wnt expression in extraocular and limb muscles from mouse models with ALS. *Vahid Harandi, Linda Mcloon, Fatima Pedrosa Domellöf, Jingxia Liu*

PS4-403 / #477 - Importance of peripheral AAV9 mediated SMN expression for gene therapy of spinal muscular atrophy mice. *Aurore Besse, Marianne Roda, Stéphanie Astord, Thibaut Marais, Martine Barkats*

PS4-404 / #526 - Homozygous mutation in Atlastin GTPase 1 causes recessive hereditary spastic paraplegia. *Sebahattin Cirak, Raul Heredia, Thomas Voit, Eric Hoffman*

PS4-406 / #48 - Immunohistochemical studies of hepatocyte growth factor in the skin of the patients with amyotrophic lateral sclerosis. *Seiitsu Ono, Mikio Fujikura, Hiroyuki Fukasawa, Kazuhiro Higashida, Tomomi Tsukie, Yoshihiko Oketa, Hiroaki Ishikawa, Kanako Yasui, Makoto Nomura, Hirotugu Mikami, Megumi Suzuki*

PS4-407 / #136 - Detection of LRP4 antibodies in serum and CSF from amyotrophic lateral sclerosis patients. *John Tzartos, Paraskevi Zisimopoulou, Michael Rentzos, Nikos Karandreas, Vasiliki Zouvelou, Panagiota Evangelakou, Anastasios Tsonis, Thomas Thomaidis, Giuseppe Lauria, Francesca Andreetta, Renato Mantegazza, Socrates Tzartos*

PS4-408 / #160 - Relevance of the disease progression pattern in amyotrophic lateral sclerosis. *Nuria Álvarez, Miguel Áng Rubio, Jordi Pascual-Calvet, Jaume Roquer*

PS4-409 / #341 - ALS onset and propagation: Insight from pulmonary function test. *Dong-Gun Kim, Kee Hong Park, Sung-Yeon Sohn, Ji-sun Kim, Yoon Ho Hong, Sung Min Kim, Kwang-Woo Lee, Kyoung suk Park, Jung Joon Sung*

PS4-410 / #380 - Home-based multidisciplinary care for ALS/MND in Moscow and Russia. *Lev Brylev, Marina Byalik, Alexandr Chervyakov, Ekaterina Dikhter, Vera Fominykh, Margarita Fominykh, Maria Ivanova, Elena Lysogorskaia, Oxana Orlova, Vasiliy Shtabnitskiy, Anna Sonkina, Alexei Vasiliev, Anna Vorobyeva, Maria Zakharova*

PS4-411 / #428 - Novel data from BENEFIT-ALS: Blinded Evaluation of Neuromuscular Effects and Functional Improvement with Tirasemtiv in patients with Amyotrophic Lateral Sclerosis. *Jeremy Shefner, Andrew Wolff, Lisa Meng, Jacqueline Lee, Joyce James, Jinsy Andrews*

PS4-412 / #432 - Does concurrent medication affect survival in amyotrophic lateral sclerosis? *Hakan Cetin, Berthold Reichardt, Judith Füzi, Gudrun Zulehner, Uros Klickovic, Jakob Rath, Michael Hagmann, Fritz Zimprich*

PS4-414 / #508 - What is the difference between brachial amyotrophic diplegia and upper limb onset ALS? Clinical and neurophysiological manifestations. *Byung-Nam Yoon, Jung-Joon Sung, Gwang-Woo Lee, Yoon-Ho Hong, Ji-Sun Kim*

PS4-416 / #61 - Muscle-intrinsic defects and atrophy both contribute to the reduction in skeletal muscle size in mouse models of spinal muscular atrophy. *Rashmi Kothary, Marc-Olivi Deguise, Justin Boyer*

PS4-418 / #137 - Amelioration of spinal muscular atrophy using RNA therapy to increase SMN level and modulate other secondary therapeutic targets. *Monica Nizzardo, Chiara Simone, Federica Rizzo, Margherita Ruggieri, Sabrina Salani, Andrea DalMas, Monica Bucchia, Emanuele Frattini, Giulia Stuppia, Giulietta Riboldi, Francesca Magri, Nereo Bresolin, Franco Pagani, Giacomo Comi, Stefania Corti*

PS4-419 / #139 - Improvement of SMARD1 phenotype using iPSC-derived neural stem cells transplantation. *Stefania Corti, Chiara Simone, Monica Nizzardo, Federica Rizzo, Margherita Ruggieri, Sabrina Salani, Monica Bucchia, Paola Rinchetti, Chiara Zanetta, Irene Faravelli, Francesca Magri, Nereo Bresolin, Giacomo Comi*

PS4-421 / #180 - Efficient SMN rescue following tricyclo-DNA antisense oligonucleotides treatment. *Valerie Robin, Aurélie Goyenvalle, Graziella Griffith, Branislav Dugovic, Christian J. Leumann, Luis Garcia*

PS4-422 / #228 - Bone Health Determinants in Spinal Muscular Atrophy (SMA) type II/III. *Natascia Di Iorgi, Giorgia Brigati, Irene Olivieri, Marta Ferretti, Marina Pedemonte, Carlo Minetti, Claudio Bruno, Mohamad Maghnie*

PS4-423 / #312 - Functional consequences of spinal muscular atrophy at the neuromuscular junction. *Anuja Neve, Tilman Voigt, Smita Saxena, Daniel Schuemperli*

PS4-425 / #355 - Identifying small molecules targeting an RNA stem-loop involved in the alternative splicing of the SMN2 gene: a therapeutic target in SMA. *Amparo Garcia-Lopez, Gianpaolo Chiriano, Ruben Artero, Leonardo Scapozza*

PS4-426 / #389 - Muscle-resident stem cells in Spinal Muscular Atrophy. *Nathalie Didier, Maria-Grazia Biferi, Thibault Marais, Giovanna Marazzi, David Sassoon, Martine Barkats*

PS4-427 / #393 - Correlation between genotype and phenotype in Algerian patients with spinal muscular atrophy. *Karima Sifi, Yamina Sifi, Nouredine Abadi, Abdelmadjid Hamri, Cherifa Benlatreche*

PS4-428 / #472 - A novel lysophospholipid inborn error of metabolism underlying a cause of a complex distal spinal muscle atrophy: neuropathy target esterase gene and its connections with organophosphorus-induced neuropathy. *Charles Lourenço, Claudia Sobreira, Stephan Zuchner, Wilson Marques Jr*

PS4-429 / #52 - Through myotubes normalization, CYTOO 2D+ increases sensitivity of Muscle Damage HCS assay. *Yoran Margaron, Mathieu Fernandes, Delphine Morales, Sébastien Degot, Alexandra Fuschs*

PS4-430 / #204 - Serum biomarkers for Duchenne Muscular Dystrophy and muscular dystrophy animal models. *Jérémy Rouillon, Aleksandar Zocevic, Thibaut Léger, Jean-Michel Camadro, Laurent Servais, Thomas Voit, Fedor Svinartchouk*

PS4-432 / #233 - A novel tool for fast, precise, interactive segmentation of skeletal muscle NMR images. *Alexey Shukelovich, Pierre-Yves Baudin, Noura Azzabou, Jean-Marc Boisserie, Julien Le Louër, Pierre G Carlier*

PS4-433 / #252 - Quantitative MRI in hypokalaemic periodic paralysis reveals age-dependent fat infiltration of lower limb muscles. *J.M. Morrow, E. Rawah, C.D.J. Sinclair, M.R.B. Evans, S. Shah, M.G. Hanna, M.M. Reilly, J.S. Thornton, T.A. Yousry*

PS4-434 / #255 - Lower limb muscle MRI findings in X-linked Charcot-Marie-Tooth disease. *A.L. Pelayo-Negro, M.R.B. Evans, J.M. Morrow, S. Shah, A.S. Carr, M.G. Hanna, T.A. Yousry, M.M. Reilly*

PS4-435 / #274 - MRI appearance of conduction block in vasculitic neuropathy. *Michael Fu*

PS4-436 / #282 - Whole body MRI study in 28 genetically confirmed Chilean patients with dysferlinopathy. *Jorge Diaz, Lisanne Woudt, Claudia Castiglioni, Jorge A. Bevilacqua*

PS4-437 / #296 - Development of a multifaceted biomarker strategy to support clinical development of utrophin modulators for Duchenne muscular dystrophy therapy. *Jon Tinsley, Francis Wilson, Graeme Horne*

PS4-438 / #342 - Diagnostic algorithm for myoadenylate deaminase deficiency based on metabolic exercise testing parameters: a prospective study. *Fabrice Rannou, Arnaud Uguen, Virginie Scotet, Cédric Le Maréchal, Odile Rigal, Pascale Marcorelles, Eric Gobin, Jean-Luc Carré, Fabien Zagnoli, Marie-Agnès Giroux-Metges*

PS4-440 / #375 - Automated tract based analysis of diffusion properties in amyotrophic lateral sclerosis. *Valeriu Culea, Florian Borsodi, Christian Langkammer, Lukas Pirpamer, Christian Enzinger, Reinhold Schmidt, Franz Fazekas, Stefan Ropele*

PS4-441 / #482 - Effect size of quantitative muscle imaging in Duchenne muscular dystrophy exceeds the effect size of clinical scores of muscle function. *Hafner Patricia, Ulrike Bonati, Andrea Klein, Cornelia Neuhaus, Oliver Bieri, Monika Gloor, Arne Fischmann, Dirk Fischer*

PS4-442 / #536 - Involvement of the brachial plexus - a combined diagnostic approach. *Stefan Meng, Manfred Frey, Anna Grisold, Wolfgang Grisold*

PS4-443 / #100 - Effect of mitochondrial changes on myopathies clinical course.
Vladimir S Sukhorukov, Dmitry A. Kharlamov, Tatiana I. Baranich

PS4-444 / #39 - Mirror movements in amyotrophic lateral sclerosis. *Mohamed Hassan, Marwa Hassan, Mohamed Hamdy, Reinhard Dengler*

PS4-445 / #97 - The reproducibility and usefulness of motor unit number index (MUNIX) using abductor digiti minimi and tibialis anterior muscles in ALS patients. *Je-Young Shin, Dong-Gun Kim, Kee Hong Park, Sung-Yeon Sohn, Ji-Sun Kim, Yoon-Ho Hong, Jung-Joon Sung, Kwang-Woo Lee*

PS4-446 / #127 - Motor unit number index (MUNIX) in the orbicularis oculi muscle of healthy subjects. *Suk-Won Ahn, Yoon-Ho Hong, Da-Eun Jeong, Ji-Won Yang, Ji-Sun Kim, Kee Hong Park, Je-Young Shin, Sung-Yeon Sohn, Jung-Joon Sung, Byung-Nam Yoon, Dong-gun Kim*

PS4-447 / #542 - Prognostic significance of A-waves as an isolated abnormity of nerve conduction studies. *Eva Vlckova, Josef Bednarik*

PS4-448 / #108 - Sequence capture and targeted resequencing in DNA diagnostics of neuromuscular diseases. *Kristyna Stehlikova, Daniela Skalova, Lenka Mrazova, Petr Vondracek, Jiri Fajkus, Lenka Fajkusova*

PS4-449 / #130 - A 7-gene signature allows the identification of altered muscle tissue.
Pia Bernasconi, Dimos Kapetis, Cristina Cappelletti, Lucia Morandi, Lorenzo Maggi, Fulvio Baggi, Francesca Zolezzi, Fabio Stella, Renato Mantegazza

PS4-450 / #178 - An amplicon-based massive parallel sequencing for diagnosis of Duchenne and Becker muscular dystrophies. *Mélissa Alame, Reda Zenagui, Delphine Thorel, Déborah Mechin, Fabienne Danton, Mireille Claustres, Michel Koenig, Mireille Cossee*

PS4-451 / #230 - Free radical oxidation in hereditary motor-sensory neuropathies and myotonic dystrophy. *Elena Saifullina, Rim Magzhanov, Rafagat Farkhutdinov*

PS4-452 / #251 - Developing Diagnostic Techniques for Dysferlinopathy. *Elaine Lee, Arunkanth Ankala, Babi Nallamilli, Esther Hwang, Madhuri Hegde, Laura Rufibach*

PS4-453 / #301 - Urine biomarkers for Duchenne Muscular Dystrophy and dystrophin-deficient animal models. *Jérémy Rouillon, Aleksandar Zocevic, Thibaut Léger, Camille Garcia, Jean-Michel Camadro, Laurent Servais, Thomas Voit, Fedor Svinartchouk*

PS4-454 / #306 - Targeted NGS sequencing using HaloPlex - for analysis of highly heterogeneous CMT neuropathy patients detected the causing gene in 24% of examined patients.
Dana Safka Brozkova, Marcela Kr?tová, Petra Lassuthova, Pavel Seeman

PS4-456 / #315 - PFKM gene defect and glycogen storage disease GSD VII with misleading histochemical activity result. *Satu Sandell, Mari Auranen, Sanna Huovinen, Anders Paetau, Päivi Piirilä, Kati Viitaniemi, Johanna Palmio, Sini Penttilä, Bjarne Udd*

PS4-457 / #365 - A large screening of myopathic patients by a targeted NGS approach reveals great genetic heterogeneity and "multiple troubles".
Marco Savarese, Giuseppina Di Fruscio, Annalaura Torella, Arcomaria Garofalo, Teresa Giugliano, Chiara Fiorillo, Giorgio Tasca, Cristina Pisano, Francesca Del Vecchio Blanco, Giulio Piluso, Olimpia Musumeci, Marina Mora, Lucia Morandi, Enzo Ricci, Tiziana Mongini, Luisa Politano, Corrado Angelini, Giacomo Pietro Comi, Claudio Bruno, Vincenzo Nigro

PS4-458 / #384 - Improving diagnostic cell-based assays for myasthenia gravis.
Saif Huda, Inga Koneczny, Leslie Jacobsen, David Beeson, Angela Vincent

PS4-459 / #458 - Array CGH in the diagnosis of neuromuscular disorders – The NMD-Chip experience in Hungary. *Veronika Karcagi, Beata Dudas, Henriett Piko, Agnes Herczegfalvi, Rita Horvath, Hanns Lochmüller, Nicolas Lévy*

PS4-460 / #506 - FGF21: A biomarker of neuromuscular diseases ?
Endre Pál, Emese Lovadi, Ágnes Seb?k, Sámuel Komoly

PS4-461 / #512 - Will the next generation sequencing strategy change deeply the diagnosis of Charcot-Marie-Tooth disease ? *Anne-Sophie Lia, Corinne Magdelaine, Marion Lafere, Hélène Dzugan-Beauvais, Jean-Michel Vallat, Franck Sturtz, Benoît Funalot*

PS4-462 / #125 - Making a positive difference for families living with a neuromuscular condition - the Muscular Dystrophy Association of New Zealand's Fieldwork Practice Framework. *Miriam Rodrigues, Chris Higgins*

PS4-463 / #132 - Actual condition survey for solitudinous patients with subacutemyelo-optico-neuropathy in Japan. *Hiroto Takada, Kaori Odaira, Shuji Hashimoto, Masaaki Konagaya*

PS4-464 / #74 - Parents' experience of having a child with spinal muscular atrophy type 1 - informing clinical practice. *Robin Forbes, Emily Higgs, Belinda McLaren, Margaret Sahhar, Monique Ryan*

PS4-465 / #154 - Construction of a disease-specific health-related quality of life scale for patients suffering from slowly progressive neuromuscular disease. *Antoine Dany, François Boyer, Damien Jolly, Moustapha Drame, Isabella Morrone, Jean-Luc Novella, Aurore Wolak-Thierry, Coralie Barbe*

PS4-466 / #200 - Experience of patients receiving their diagnosis of myotonic dystrophy as compared with Huntington's disease.
Richard Roxburgh, David Bourke, Jo Dysart, Miriam Rodrigues

PS4-467 / #412 - Gender differences in predictors for social network and quality of life in DM1 patients. *Gro Solbakken, Torunn Dahl Eikeland, Tormod Hagen, Terje Nærland*

PS4-468 / #241 - Effect of ankle-foot orthosis on the gait biomechanics of a patient with Duchenne muscular dystrophy. *Mariana Souza, Cyntia Rogean de Jesus Alves de Baptista, Marisa Figueiredo, Elizângela Aparecida da Silva Lizzi, Rogério Ferreira Liporaci, Ana Cláudia Mattiello-Sverzut*

PS4-469 / #349 - Multidisciplinary respiratory care support team can reduce respiratory complications of neuromuscular disease inpatient. *Kiyonobu Komai, Atsuro Tagami, Chiho Ishida, Kazuya Takahashi, Yuko Motozaki, Ichiro Nozaki, Tokuhei Ikeda*

PS4-470 / #501 - Early management intervention and parent empowerment in neuromuscular diseases: the clinical center NEMO experience. *Ksenija Gorni, Valentina Morettini, Elisa Torretta, Cristina Grandi, Viviana Baiardi, Valeria Sansone*

PS4-471 / #510 - Hydrotherapy program in Duchenne Muscular Dystrophy : motor functional evaluation and body self perception. *Ksenija Gorni, Cristina Grandi, Gabriella Giuliano, Valentina Morettini, Viviana Baiardi, Valeria Sansone*

PS4-472 / #69 - Duchenne muscular dystrophy- a correlational study of upper limb performance with ambulatory performance. *Joy Goubran, Monique Ryan*

PS4-473 / #89 - Rasch Analysis of the Motor Function Measure in Patients with Congenital Muscle Dystrophy and Congenital Myopathy. *Carole Vuillerot, Pascal Rippert, Virginie Kinet, Anne Renders, Mina Jain, Melissa Waite, Allan M Glanzman, Françoise Girardot, Dalil Hamroun, Jean Iwaz, René Ecochard, Carole Bérard, Isabelle Poirot, Carsten G Bönnemann*

PS4-474 / #91 - English cross-cultural translation and validation of the NM-Score: a system for motor function classification in patients with neuromuscular diseases. *Carole Vuillerot, Katherine Meilleur, Mina Jain, Melissa Waite, Tianxia Wu, Jahannaz Datsgir, Sandra Donkervoort, Meganne Leach, Anne Rutkowski, Pascal Rippert, Christine Payan, Jean Iwaz, Dalil Hamroun, Carole Bérard, Isabelle Poirot, René Ecochard, Carsten G Bönnemann*

PS4-475 / #209 - 1000 Norms Project: clinical catalogue of human neuromuscular variation.
Marnee McKay, Jennifer Baldwin, Milena Simic, Paulo Ferreira, Niamh Moloney, Claire Hiller, Jean Nightingale, Joshua Burns

PS4-476 / #237 - Oxygen uptake evaluated with two different methods using upper limbs of the non-ambulatory children. *Marisa Figueiredo, Monalisa Squiaveto, Luciano Oliveira, Mariana Souza, Lourenço Gallo, Ana Cláudia Mattiello-Sverzut*

PS4-477 / #253 - Cross-Cultural Adaptation, Reliability, And Validity Of The Turkish Version Of Motor Function Measure (MFM-TR). *H.Serap Inal, Ela Tarakci, Gülcan Aksoy, Sezen Mergen K?l?c, Hakan Beser, Cigdem Beser, Yesim Gulsen Parman, Feza Deymeer, Piraye Oflazolu*

PS4-478 / #263 - Foot deformity and stabilometric parameters in children with charcot-marie-tooth disease. *Amanda Testa, Tais Regina Silva, Cyntia Alves de Baptista, Wilson Marques Junior, Ana Cláudia Mattiello-Sverzut*

PS4-479 / #368 - The NorthStar ambulatory assessment in Duchenne muscular dystrophy: considerations for the design of clinical trials. *Valeria Ricotti, Deborah Ridout, Marika Pane, Ros Quinlivan, Stephanie Robb, Eugenio Mercuri, Adnan Manzur, Francesco Muntoni*

PS4-480 / #418 - Transcutaneous capnography as early indicator of nocturnal hypoventilation in neuromuscular disorders. *Federica Trucco, Marina Pedemonte, Chiara Fiorillo, Claudio Bruno, Carlo Minetti*

PS4-481 / #518 - Long-term follow-up of sporadic Inclusion Body Myositis using dynamometry. *Jean-Yves Hogrel, Yves Allenbach, Aurélie Canal, Gaelle Leroux, Gwenn Ollivier, Kuberaka Mariampillai, Laurent Servais, Serge Herson, Valérie Decostre, Olivier Benveniste*

PS4-482 / #522 - One-year follow-up of patients with Duchenne muscular dystrophy using high precision tools for upper limb assessment. *Jean-Yves Hogrel, Andrea Seferian, Amélie Moraux, Mélanie Annoussamy, Aurélie Canal, Valérie Decostre, Oumar Diabate, Anne-Gaelle Le Moing, Teresa Gidaro, Nicolas Deconinck, Frauke Van Parys, Wendy Vereecke, Sylvia Wittevrongel, Michèle Mayer, Kim Maincent, Isabelle Desguerre, Christine Themar-Noel, Jean-Marie Cuisset, Vincent Tiffereau, Séverine Denis, Virginie Jousten, Thomas Voit, Laurent Servais*

PS4-483 / #530 - GNE Myopathy Functional Activity Scale (GNEM-FAS): Results from a Phase 2 study of extended release sialic acid (SA-ER). *Zohar Argov, Faye Bronstein, Yoseph Caraco, Alicia Esposito, Yael Feinsod-Meiri, Juliane Florence, Eileen Fowler, Marcia Greenberg, Edwin Kolodny, Heather Lau, Alan Pestronk, Odelia Rebibo, Perry Shieh, Catherine Siener, Elizabeth Malkus, Jill E. Mayhew, Alison Skrinar*

PS4-484 / #545 - Evaluation of dysphagia in GRMD dogs using respiratory inductance plethysmography. *Inès Barthélémy, Xavier Cauchois, Isabel Punzon, Jean-Laurent Thibaud, Stéphane Blot*

PS4-485 / #546 - Comparative respiratory function evaluation in two canine myopathies using respiratory inductance plethysmography. *Inès Barthélémy, Xavier Cauchois, Isabel Punzon, Jean-Laurent Thibaud, Stéphane Blot*

PS4-486 / #288 - THE New Zealand Neuromuscular Disease Registry – A review of diagnoses confirmed by molecular test.
Miriam Rodrigues, Alexa Kidd, Donald Love, Richard Roxburgh

PS4-487 / #323 - Serum proteins as predictive biomarkers of rAAV efficiency in translational studies. *Jérôme Denard, Christine Jenny, Thibaut Léger, Camille Garcia, Jean-Michel Camadro, Thomas Voit, Fedor Svinartchouk*

PS4-488 / #343 - Changes of deep paraspinal muscles in idiopathic scoliosis: a pilot electrophysiological and histochemical study. *Josef Zamecnik, Ivana Stetkarova, Jaromir Hacek, Robert Artur Dahmen, Martin Krbec*

PS4-489 / #347 - Clinical manifestation and disease course of the patient with HAM/TSP. *Eiji Matsuura, Satoshi Nozuma, Osamu Watanabe, Hiroshi Takashima*

PS4-490 / #373 - Exploring the SOD1-G93A transgenic swine as a novel animal model for Amyotrophic Lateral Sclerosis (ALS). *Cristiano Corona, Paola Crociara, Caterina Bendotti, Alberto Botter, Maria Novella Chieppa, Antonio D'Angelo, Roberto Duchi, Donato Formicola, Monica Lo Faro, Roberto Merletti, Alberto Rainoldi, Cesare Galli, Cristina Casalone*

PS4-491 / #396 - Neuromuscular complications of HTLV-I: Case Reports. *Reza Boostani*

PS4-492 / #397 - Patterns on spinal magnetic resonance image in pediatric Guillain-Barré syndrome. *María Sol Cormick, Juan Pablo Princich, Carlos Rugilo*

PS4-493 / #420 - Systemic diseases diagnosed by muscle biopsy. *Adrián Téllez, José C. Milisenda, Ricardo A. Losno, Alba Jerez, María D. Cano, Josep M. Grau*

PS4-494 / #476 - Pronoctic Features of LGMD presenting as symptomatic or pacisymptomatic HyperCKemia. *Pilar Martí, Nuria Muelas, OIHANE JAKA, Amets Sáenz, Fernando Mayordomo, Inmaculada Azorín, Pia Gallano, Adolfo López-de munían, JJ Vilchez*

PS4-495 / #491 - A Rare Reason For Paraplegias: Myositis Ossificans. nebahat tasdemir, *Nebahat Tasdemir*

PS4-496 / #496 - Multisegmental Anterior Horn Motor Nueron Disease Due To A Case. *Nebahat Tasdemir*

PS4-497 / #499 - AFM-Téléthon / Bpifrance Seed Fund dedicated to innovative biotherapies and rare diseases. *Jean-Pierre Gaspard*

LUNCH SYMPOSIUM

MONDAY JULY 7, 2014

12:30-14:00 **GENZYME SYMPOSIUM** (ROOM HERMES)

PATHOLOGIC, THERAPEUTIC AND DIAGNOSTIC ADVANCES IN POMPE DISEASE.
Chair: Claude Desnuelle (France)

12.30 **Welcome and Introduction.** *Claude Desnuelle (France)*
12.40 **Autophagy as a pathophysiological and therapeutic approach.** *Marco Sandri (Italy)*
13.00 **The effect of alglucosidase Alpha on muscle biopsy features of Pompe disease.** *Maurizzio Moggio (Italy)*
13.15 **The obstacles towards suspecting Pompe disease.** *Alberto Dubrovsky (Argentina)*
13.35 **The systematic approach to the diagnosis of myopathies.** *Tracey Willis (UK)*
13.55 **Closing remarks.** *Claude Desnuelle (France)*

TUESDAY JULY 8, 2014

12:30-14:00 **AFM-TELETHON SYMPOSIUM** (ROOM HERMES)
Chairmen: Thomas Voit (France), Serge Braun (France)

3D imaging of SMA (and other indications). *Linda Lowes (USA)*
Upper limb outcome measure tools. *Eugenio Mercuri (Italy)*
Continuous activity monitoring of ambulant and non-ambulant patients. *Jean-Yves Hogrel (France)*
Innovative methods for assessing the neuromuscular function in non-ambulant patients. *Jean-Yves Hogrel (France)*

WEDNESDAY JULY 9, 2014

12:30-14:00 **PFIZER SYMPOSIUM** (ROOM CLIO)

TTR-FAP – AVOIDING MISDIAGNOSIS OF A RARE POLYNEUROPATHY
Chair: Teresa Coelho (France)

12:30 **Introduction.** *Teresa Coelho (France)*
12:35 **TTR-FAP – the rare disease that specialists need to know about.** *Teresa Coelho (France)*
12:50 **Clinical scenarios – avoiding the misdiagnosis of complex polyneuropathy.**
Lucía Galan (Spain), Andoni Echaniz-Laguna (France), Isabel Illa (Spain)
13:40 **TTR-FAP in the clinic.** *Teresa Coelho (France)*
14:00 **Close**

12:30-14:00 **SAREPTA SYMPOSIUM** (ROOM CALLIOPE)

Exon Skipping: A Disease-Modifying Platform in Development to Treat DMD
Chair: Ed Kaye (USA)

- *Anne Connolly (USA)*
- *Eugenio Mercuri (Italy)*
- *Francesco Muntoni (UK)*
- *Thomas Voit (France)*

THURSDAY JULY 10, 2014

12:30-14:00 **PTC THERAPEUTICS SYMPOSIUM** (ROOM CALLIOPE)

New perspectives on the diagnosis and management of Duchenne muscular dystrophy
Chair: Francesco Muntoni (UK)

12:30 **Welcome and introduction.** *Francesco Muntoni (UK)*
12:40 **Diagnosing DMD in the era of mutation-specific therapies.** *Annemieke Aartsma-Rus (The Netherlands)*
13:00 **What is a realistic target therapeutic benefit for DMD patients and their families ?** *Eugenio Mercuri (Italy)*
13:30 **Understanding long-term outcomes in DMD.** *Hanns Lochmüller (UK)*
13:50 **Questions and meeting close.** *Francesco Muntoni (UK)*

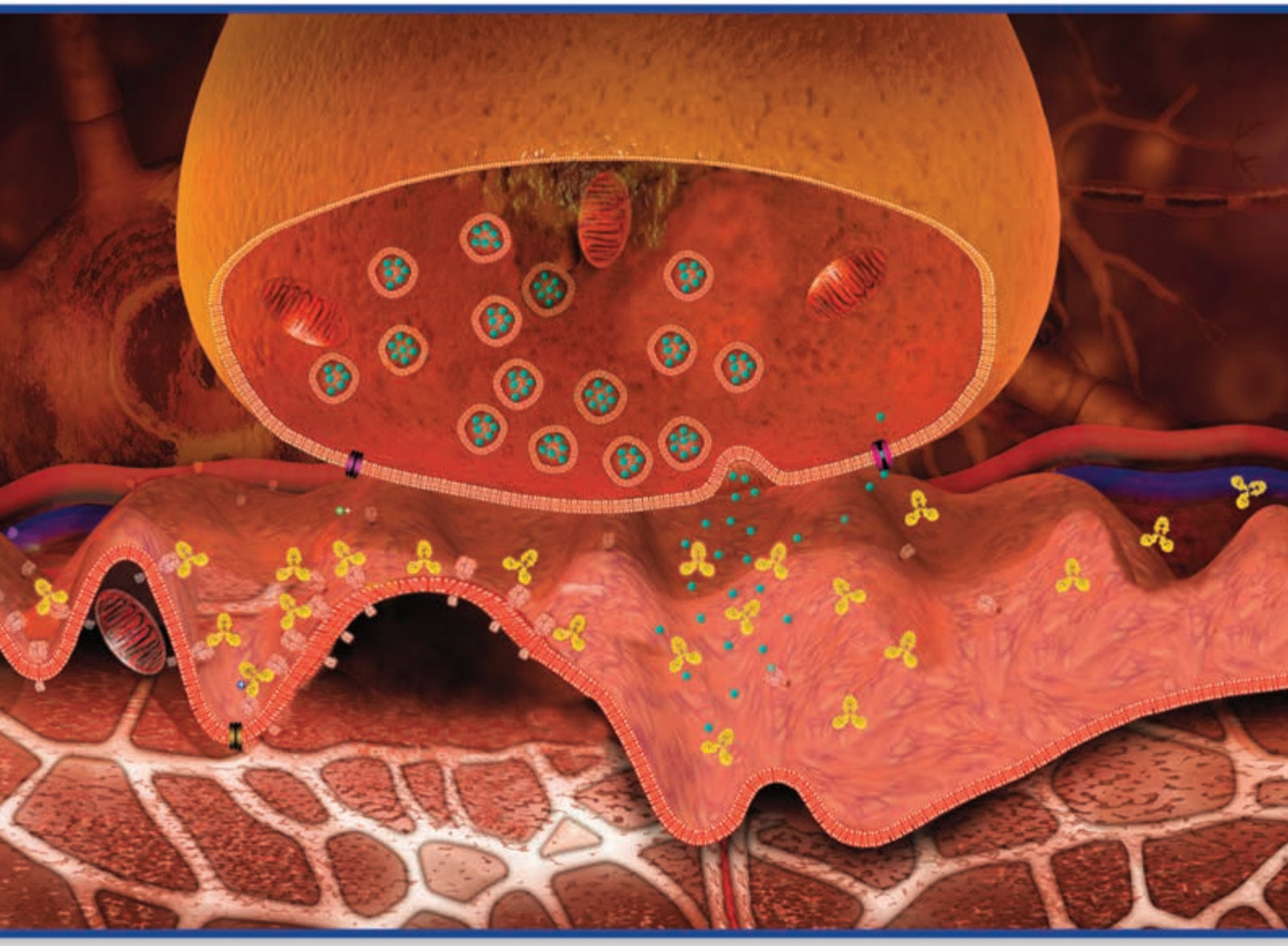
EXHIBITION MAP

EXHIBITORS

BOOTH NR

ALEXION 13
BIOMARIN .....4 & 20
CEVIDRA 1
CYTOO 15
GENEA BIOCELLS 8
GENZYME 12
LFB BIOMEDICAMENTS 16
OCTAPHARMA 21
PFIZER 11
PTC THERAPEUTICS 2
SAREPTA 6
TERUMO 9
ULTRAGENYX PHARMACEUTICAL 22

ALEXION



REFRACTORY MYASTHENIA GRAVIS:
an autoimmune disorder of neuromuscular transmission

For more information, visit Alexion booth n° 13

GENERAL INFORMATION

CONGRESS VENUE

Nice Acropolis, Esplanade Kennedy, 06302 Nice cedex 4, France

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Event ID : 17950AF
Validity dates : from 30/06/14 to 16/07/14
Event location: Nice, France

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LFB
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PTC0609NM001
Prepared June 2014

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Genzyme exhibition booth #12

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genzyme
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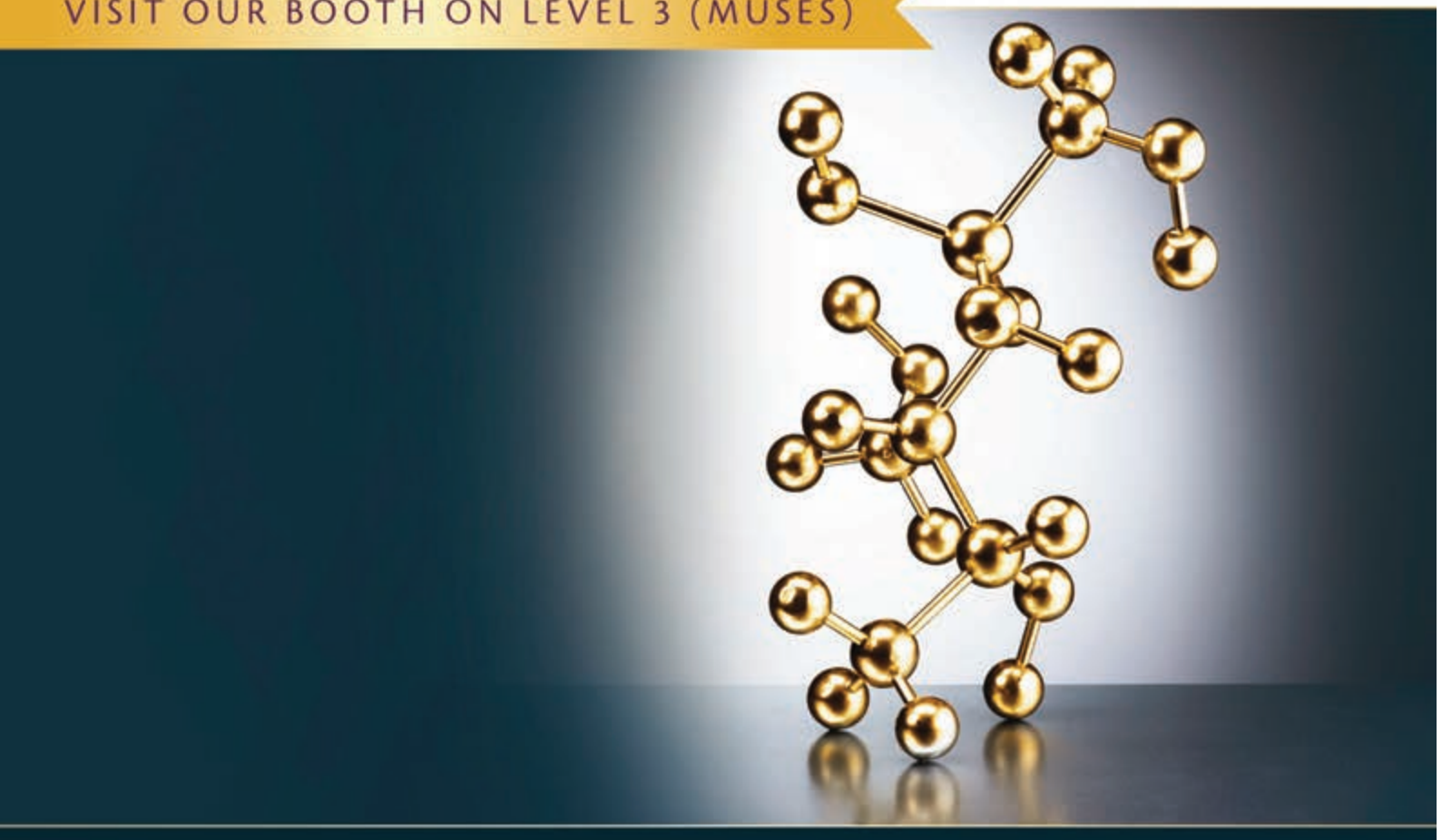
Date of preparation: June 2014
GZEMEA.PD.14.06.0049a

SAREPTA THERAPEUTICS

TRANSFORMATION, WITHIN REACH

VISIT OUR BOOTH ON LEVEL 3 (MUSES)



We are Sarepta Therapeutics. Our goal is to develop first-in-class, RNA-based therapeutics to improve the lives of people affected by serious and life-threatening rare genetic disorders.

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We invite you to join our symposium

EXON SKIPPING: A DISEASE-MODIFYING PLATFORM IN DEVELOPMENT TO TREAT DMD

Wednesday, July 9th, 12:30 – 14:00

Calliope Room | Level 3 (Muses)

Lunch will be served at 12:00

Please see our flyer in your registration bag for a detailed agenda.

DMD-00038-1-06014