World Federation of Neurology

XII International Congress on Neuromuscular Diseases

in Memory of Lucia Ines Comi

Naples, Italy

July 17th-22nd, 2010

PROGRAM
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Giovanni Nigro

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Luisa Politano, Simone Sampaolo, Cira Solimene

Congress Secretariat
Congrex Group, Göteborg, Sweden
Welcome to XII International Congress on Neuromuscular Diseases

On behalf of the Myology groups of the Second University of Naples, and the Veterinary Medical School of Federico II Naples University, I welcome the participants in the XII International Congress on Neuromuscular Diseases. My co-workers and I are very grateful to the research group on neuromuscular disorders of the World Federation of Neurology, who selected Naples for this important project. Furthermore we would thank both Naples Universities, Gaetano Conte Academy, Mediterranean Society of Myology, the Congrex Agency from Göteborg and Perrotta catering from Naples for their help with Congress organization. We are particularly grateful to Valerie Askanas, President of the Scientific Committee, and to all other members of her Committee for the interesting and innovating scientific programme. Everyday, step by step, the field of neuromuscular disorders becomes greater, and it is now ranging from Genetics to Immunology, from Neurology to Cardiology, from Pathology to Social Medicine, from Radiology to Veterinary Myology, from Respiratory Care to Orthopaedic Surgery and Physical/Rehabilitation Medicine. Following these statements, the Congress will consider the new aspects of the neuromuscular disorders, giving the most recent information on each aspect of Neuro-Myology. Finally, we are happy that so many friends may have the opportunity to enjoy the natural beauties of our district, visiting at least a part of the vast amount of cultural gems spanning 25 centuries of life of Naples and surroundings. Therefore we suggest you to prolong your stay in Naples to see at least Capri and Pompei, the old city of Naples and the other beautiful places that Goethe named “Arcadia”.

Giovanni Nigro
President of the XII International Congress on Neuromuscular Diseases

Greetings from the President of the Scientific Program Committee

Dear Colleagues,

On behalf of the Scientific Program Committee of the XII International Congress on Neuromuscular Diseases, I warmly welcome you to the Congress and to beautiful Naples. Knowing that the Congress’ Participants will have different requirements, backgrounds, and interests, we have tried to organize an interesting, and scientifically diverse program. We emphasize both the basic science and clinical aspects of neuromuscular diseases. We realize that there are some topics that could not be included due to the time restriction, but we trust that the richness of the program covering the newest developments in diagnoses, pathogeneses and treatments of a wide spectrum of neuromuscular diseases will provide an enriching experience. And now, let the sessions begin!

Enjoy the Congress!

Valerie Askanas
President, Scientific Program Committee
XII International Congress on Neuromuscular Diseases
Patronages

World Federation of Neurology
President of the Republic of Italy
Prime Minister of the Republic of Italy
Ministry of Public Health
Ministry of University and Research
Ministry of Pari Opportunità

Second University of Naples (SUN)
University of Naples Federico II
Faculty of Medicine (SUN)
Department of Experimental Medicine (SUN)

Mediterranean Society of Myology
Gaetano Conte Academy
Italian Association of Myology
Italian Society of Neurology
Association Française contre les Myopathies
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Piccin Editore – Agenzia Napoli
**XII International Congress on Neuromuscular Diseases**

**PROGRAM (Summary)**

**Saturday – July 17th, 2010**

h. 8.00 – 13.00 **CONCURRENT TEACHING COURSES (TC1-TC5)**

h. 8.00 – 8.30 **Registration**

**TC1: Old and new problems in neuromuscular disorders (Hall F5)**

h. 8.30 – 10.30 **Sporadic and congenital Myasthenia: how to diagnose and treat**
H. Lochmüller (UK)

**Diabetic and other dysimmune neuropathies: pathogenesis and treatment options**
W. King Engel (USA)

h. 10.30 – 11.00 **Coffee break**

h. 11.00 – 13.00 **Metabolic Myopathies: new phenotypes and treatable conditions**
C. Angelini (Italy)

**Neuromuscular Disorders in infancy: What’s new about phenotypes and clinical trials**
F. Muntoni (UK)

**TC2: Neuromuscular immunology for clinicians (Hall F1)**

h. 8.30 – 10.30 **Practical basic immunology for neuromuscular specialists, including evaluation of relevant therapeutic agents**
M. Dalakas (UK)

**Peripheral Neuropathies an immunologic approach to diagnosis**
C. Briani (Italy)

h. 10.30 – 11.00 **Coffee break**

h. 11.00 – 13.00 **Inflammatory Myopathies: Practical aspects of diagnosis and treatment**
F. Mastaglia (Australia)

**Antibodies in Myasthenia: how to diagnose them**
A. Vincent (UK)

**TC3: Genetics for clinicians (Hall F2)**

h. 8.30 – 10.30 **How important is a precise diagnosis for patients with muscular dystrophies and other myopathies**
V. Nigro (Italy)

**The quaint genetic behaviour in mitochondrial myopathies**
S. Di Mauro (USA)

h. 10.30 – 11.00 **Coffee break**

h. 11.00 – 13.00 **Trinucleotide Repeat Disorders: the pathogenic basis of clinical manifestations. Possible therapeutic approaches**
G. Novelli (Italy)
Genetic Neuropathies: Is categorizing important? Treatment, now or in near-future?
M. Reilly (UK)

TC4: Clinical, histochemical and electrophysiologic approaches to neuromuscular diseases (Hall F3)

h. 8.30 – 10.30  Clinical clues in the diagnosis of adult hereditary myopathies
Z. Argov (Israel)
Histochemical and electron-microscopic approach to muscle diseases: slide sessions
A. Oldfors (Sweden)

h. 10.30 – 11.00  Coffee break

h. 11.00 – 13.00  Practical pearls from neurophysiologic experience with peripheral neuropathies
L. Santoro (Italy)
The new inherited pathological basis of motor neuron disorders
R. Baloh (USA)

TC5: Diagnosis and management of acute neuromuscular disorders (Hall F4)

h. 8.30 – 10.30  Acute peripheral nerve disorders
G. Said (France)
Management and treatment of the myasthenic crisis
A. Evoli (Italy)

h. 10.30 – 11.00  Coffee break

h. 11.00 – 13.00  Rhabdomyolysis: causes and possible therapies
A. Toscano (Italy)
How to deal with sudden muscle weakness
M. Hanna (UK)

h. 18.30  OPENING CEREMONY (ROYAL PALACE THEATRE)
Chairs: Reinhardt Rüdel, Gerard Said, Gioacchino Tedeschi

Addresses

President of the World Federation of Neurology
President of the Scientific Program Committee
Rector of the University of Naples Federico II
Rector of the Second University of Naples
President of the Congress

Honorary Lectures

Alan E.H. Emery
Lord John Walton of Detchant

Welcome Dinner – Naples Royal Palace
Sunday – July 18th, 2010

h. 8.00 – 18.00 Registration
h. 8.00 – 10.00 Setting up Posters (SM), coffee and rolls

h. 10.00 – 12.00 PLENARY LECTURES (Red and Blue Halls)
Chairs: Giovanni Nigro (Italy) and W. King Engel (USA)

h. 10.00 – 10.30 PL1. Recent advances in understanding pathogenesis and treatment of Mitochondrial Myopathies
Salvatore Di Mauro (USA)

h. 10.30 – 11.00 PL2. State of the art in muscle glycogenosis
Corrado Angelini (IT)

h. 11.00 – 11.30 PL3. Inclusion-Body Myositis: newest pathogenic findings paving the way to possible treatments
Valerie Askanas (USA)

h. 11.30 – 12.00 PL4. Clinical and serological subgroups of Myasthenia Gravis and their relevance to formulating treatment
Angela Vincent (UK)

h. 12.15 – 13.30 Lunch (self-served buffet)

h. 13.30 – 15.00 GENZYME SYMPOSIUM (Red Hall)

h. 15.00 – 16.00 MEET THE PROFESSOR – 2 CONCURRENT SESSIONS

MTP1 (Hall E1) Cardiac and Respiratory involvement in Neuro-Muscular Diseases
Giovanni Nigro (Moderator) – E. Arbustini – D. Duboc – S. Iannaccone – L. Politano

MTP2 (Red Hall) Genetic testing and its role in diagnosis
K. North (Moderator) – K. Christodoulou – V. Nigro – M. Shy

h. 16.00 – 18.00 SIMULTANEOUS SYMPOSIA (S1-S7)

S1 (Hall E1) Muscle weakness in aging
Chairs: W. King Engel and Olivier Benveniste

Invited Speakers

S1.1. Mitochondrial Dysfunction of Aging Skeletal Muscle: A potential therapeutic Focus
David A. Hood

S1.1.2. Histochemical abnormalities in aging human muscle: Which might be preventable or treatable?
W. King Engel
S1.L3. A critique of aging-animal models in the study of mechanisms underlying age-related loss of skeletal muscle mass and function
Malcolm Jackson

Platform Presentations

S1.PP1. Differential features of muscle atrophy in patients with osteoporosis and osteoarthritis
C. Terracciano, A. Gattelli, E. Lena, U. Tarantino, R. Massa

S1.PP2. Administration of Losartan improves skeletal muscle repair and modulates strength in mice with sarcopenia
T. Burks, E. Andres-Mateos, J.L. Simmers, E.M. MacDonald, C. van Erp, R. Rattner, R.D. Cohn

S2 (Hall E2) Pathogenic Mechanisms of Inherited Neuropathies
Chairs: Michel Shy and Simone Sampaolo

Invited Speakers

S2.L1. Mitochondrial fusion and transport in inherited axonal neuropathies
Robert Baloh

S2.L2. Clinical Approach to Inherited Neuropathies
Mary Reilly

S2.L3. Schwann cell pathways in inherited demyelinating neuropathies
Lawrence Wrabetz

Platform Presentations

S2.PP1. Clinical and electrophysiological characteristics of CMT4C4 disease caused by L239F mutation in the GDAP1 gene: an example of phenotype-genotype correlations

S2.PP2. Axonal degeneration precedes demyelination of peripheral nerves of mice lacking Cx32 gap junctions in the myelin sheath
I. Sargiannidou, N. Vavlitou, K. Markoullis, K. Kyriacou, S. Scherer, K. Kleopa

S2.PP3. High-throughput mutation analysis of the human aminoacyl-tRNA synthetase genes: in search of additional loci responsible for inherited peripheral neuropathies
A. Antonellis, V. Ionasescu, J. Lupski, G. Nicholson, K. Talbot, J. Vance, S. Zuchner, E. Green

S3 (Hall F1) Update on Lipid muscle diseases
Chairs: Giuseppe Vita and Generoso Andria

Invited Speakers

S3.L1 Triglycerides storage diseases
Claudio Bruno

S3.L2. Clinical and laboratory features of fatty acid disorders
Ingrid Tein
S3.L3. Myopathic forms of multiple acid CoA dehydrogenase deficiency (MADD)
Rita Horvath

Platform Presentations

S3_PP1. Modulation of autophagy as a therapeutic approach to a lysosomal storage disorder: Pompe disease
N. Raben, C. Schreiner, R. Baum, P. Plotz

S3_PP2. Late-onset multiple acyl-CoA dehydrogenase deficiency (MADD): a clinical, biochemical and genetic study in 10 patients

S4 (Hall F2)  Novel therapeutic targets at the neuromuscular junction
Chairs: Lefkos Middleton and Amelia Evoli

Invited Speakers

S4.L1. Experimental models and treatments in Congenital Myasthenic Syndromes
David Beeson

S4.L2. Gene experimental therapies in Myasthenia Gravis
Marc de Baets

S4.L3. Complement involvement in pathogenesis and treatment
Henry Kaminski

Platform Presentations

S4_PP1. Experimental anti-Muscle Specific Kinase (MuSK) myasthenia (EAMM) in lewis rats
D. Richman, K. Nishi, S. Morell, R. Maselli, M. Agius

S5 (Hall F3)  Basic mechanisms in Spinal Muscular Atrophy
Chairs: Judith Melki and Susan Iannaccone

Invited Speakers

S5.L1. The SMN complex, mechanism and therapeutic targets in spinal muscular atrophy
Gideon Dreyfuss

S5.L2. Developmental aspects of Spinal Muscular Atrophy
Eduardo F. Tizzano

S5.L3. Management of Bulbar and spinal muscular atrophy (Kennedy’s disease)
Joseph Finsterer

Platform Presentations

S5_PP1. Asymptomatic members of SMA families with homozygous mutations in the SMN1 gene
M. Jedrzejowska, M. Milewski, J. Zimowski, A. Kostera-Pruszczyk, K. Szczaluba, M. Jurek, D. Sielska, I. Hausmanowa-Petrusewicz
**S5.PP2. Autosomal dominant adult Spinal Muscular Atrophy with dysautonomia**  
R. Liguori, F. Pizza, V. Donadio, L. Caporali, M.L. Valentino, P. Avoni, V. Carelli, P. Montagna

**S5.PP3. TGFbeta impairs skeletal muscle differentiation and induces autophagy by PED/PEA-15-mediated pathway**  
S. Iovino, F. Oriente, G. Botta, S. Cabaro, F. Beguinot, P. Formisano

**S6 (Red Hall) Cardiomyopathies (in honour of Ketty Schwartz)**  
*Chairs: Giovanni Nigro and Grzegorz Opolski*

**Invited Speakers**

**S6.L1. Primary dilated cardiomyopathies**  
Eloisa Arbustini

**S6.L2. A new insight in the pathophysiology of arrhythmias in Steinert’s disease**  
Denis Duboc

**S6.L3. The alternative atrial stimulation site in Myotonic dystrophy type 1 patients**  
Gerardo Nigro

**S6.L4. Cardiac involvement in nucleopathies and its relation to skeletal muscle involvement**  
Grzegorz Opolski, Irena Hausmanowa Petrusewicz

**Platform Presentations**

**S6.PP1. Beneficial effects of angiotensin-converting enzyme inhibitors in Dystrophinopathic Cardiomyopathy**  
L. Politano, A. Palladino, L. Passamano, M.R. Cecio, A. Taglia, M.G. Di Gregorio, G. Nigro

**S6.PP2. The value of cardiovascular magnetic resonance in the follow-up of Duchenne/Becker muscular dystrophy**  

**S7 (Hall F4) Veterinary Myology**  
*Chairs: Serenella Papparella and John Mc Howell*

**Invited Speakers**

**S7.L1. Canine and feline muscular dystrophies: state of the art**  
Diane G. Shelton

**S7.L2. New advances in canine and feline inflammatory myopathies**  
Orlando Paciello

**S7.L3. Gain of function mutation in Glycogen Synthase (GYS1) causes a novel skeletal muscle glycogenosis**  
Stephanie Valberg
**Platform Presentations**

**S7.PP1. Role of the serotonergic system in the pathogenesis of fibrosis in canine idiopathic inflammatory myopathies**  
L. M. Pavone, F. Trapani, A. Spina, S. Rea, L. Avallone, S. Papparella, O. Paciello

**S7.PP2. Muscle biopsy in the diagnosis of flexural deformities (FD) in foals**  
M. P. Pasolini, L. Auletta, F. Trapani, S. Papparella, F. Lamagna, O. Paciello

**S7.PP3. Vacuolar myopathy of unknown origin in the horse**  
E. Ludvikova, Z. Lukas, P. Jahn

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**POSTER PARALLEL SESSIONS (1 – 5)**

**h. 18.00 – 19.30**

**POSTER SESSION 1:**

**MUSCLE WEAKNESS IN AGING & EXERCISE THERAPY IN NEUROMUSCULAR DISEASES**  
*Chairpersons: D. Hood, F. Deymeer*

**S1**

**SM01. Characteristics of myopathies in the elderly**  
*A. Echaniz-Laguna, M. Mohr, B. Lannes, C. Tranchant*

**SM02. Cognitive impairment as a predictor of functional disability in older Georgian population**  
*I. Verulashvili*

**SM03. Multifocal motor neuropathy accompanying with hyperreflexia and paresthesia: A case report**  
*G. Celik, S. Dener*

**SM04. Two siblings presenting paraspinal muscle atrophy (axial myopathy) with HTLV-I-associated myelopathy/tropical spastic paraparesis**  
*E. Matsuura, S. Jacobson, I. Higuchi, K. Arimura, T. Hiroshi*

**SM05. Muscle pogy in camptocormia**  
*M. X. Wang, H. J. Lee, S. Brammah, R. Pamphlett*

**S24**

**SM06. Oral exercise with hot pack in myotonic dystrophy**  
*S. Nozaki, M. Kawai, Y. Umaki, T. Kimura, T. Matsumura*

**SM07. Comparing gait performance of people with Charcot-Marie-Tooth disease who do and do not wear ankle foot orthoses**  
*G. Ramdharry, A. Pollard, J. Marsden, M. Reilly*

**SM08. Exploring the experience of living with fatigue in people with Charcot-Marie-Tooth disease - a qualitative study**  
*G. Ramdharry, M. Reilly, J. Marsden*

**SM09. A pilot study of Creatine Kinase and Homocysteine levels in amyotrophic lateral sclerosis (ALS) patients under rehabilitative treatment**  
*A. Vallelunga, M. Marchetti, R. Fiaibani, A. Marcante, I. Di Santo, A. Merico, M. Bajo, P. Tonin, A. Leon, F. Piccione*

**SM10. Resistance training in patients with LGMD2I, 2A and Becker muscular dystrophy**  
*M. L. Sveen, S. Andersen, J. Vissing, T. Krag, L. H. Ingelsrud, S. Blichter, S. Jönck*

**SM11. Physical training in boys with Duchenne Muscular Dystrophy: a protocol of the No Use is Disuse study**  
*M. Jansen, I. J. M. de Groot, N. van Alfen, A. C. H. Geurts*

**SM12. Infrared tracking for locomotion and devices control in quadriplegic people**  
*M. Plaza*
SM13. Effects of chronic exercise in Duchenne muscular dystrophy
A. Dubrovsky, J. Corderi, L. Mesa

SM14. Perception of health related quality of life in patients with hereditary and acquired neurological diseases: A retrospective clinical study
M. Kılınç, S. Aksu Yıldırým, S. Atay Yýlmaz, E. Tan

SM15. The effect of body composition on motor performance in adult patients with neuromuscular diseases
S. Aksu Yıldırým, M. E. Yağa, E. Tan

POSTER SESSION 2:
CARDIOMYOPATHIES & NUCLEAR ENVELOPE DISEASES
Chairpersons: G. Bonne, G. Nigro

S6


SM17. Cardiac abnormalities in myotonic dystrophy type 2 (DM2)
M. Cabrera Serrano, A. Jaffe, N.P. Young, J.R. Daube, M. Milone

SM18. Dilated cardiomyopathy in POMT1 mutated patients with different degrees of muscle involvement

SM19. Left ventricular diastolic function in Myotonic Dystrophy
A. Palladino, G. Nigro, F. De Luca, F. Spina, G. Nigro, L. Politano

SM20. Cardiac function in Emery-Dreifuss patients and carriers
M. Marchel, A. Madej-Pilarczyk, J. Kochanowski, I. Hausmanowa-Petrusewicz, G. Opolski

SM21. Mitochondrial biogenesis and oxidative stress in the natural history of mitochondrial cardiomyopathies

S14

SM22. Report of a family with two different hereditary diseases leading to early cardiac involvement
I. Conceição, T. Evangelista, P. Pereira, E. Vieira, J. Oliveira, R. Santos

SM23. Overlapping syndrome with predominant cardiac phenotype in patient with LMNA gene mutation
A. Madej-Pilarczyk, M. Janus, R. Wojnicz, A. Fidzianska, M. Marchel, A. Niezgoda, S. Grajek, I. Hausmanowa-Petrusewicz

SM24. The Italian network for laminopathies
G. Lattanzi

SM25. Aberrant splicing in LMNA gene caused by a novel mutation on the polypyrimidine tract of the intron 5
N. Carboni, M. Floris, A. Mateddu, G. Marrosu, E. Solla, M. Porcu, E. Cocco, M. Mura, M.A. Maioli, S. Marini, R. Piras, M.G. Marrosu

M. Milone, A. Emslie-Smith, J. Knapik, A.G. Engel

SM27. Proteomic characterization of neuromuscular patients carrying lamin A/C mutations

SM28. Skeletal and heart muscle activity by FDG uptake detected on PET in Emery-Dreifuss patients, preliminary report
M. Marchel, M. Kobylecka, A. Madej-Pilarczyk, I. Hausmanowa-Petrusewicz, L. Krolicki, G. Opolski
SM29. Screening for LMNA mutations in children with genetically unclassified lower motor unit disorders
M. Menezes, E. Barlow, R. Pedersen, H. Johnston

SM30. Clinical heterogeneity in a large family with a mutation in the LMNA gene
L. Gonzalez-Quereda, M. Olive, J. Bautista, C. Paradas, J. Juan, M.J. Rodríguez, E. Verdura, E. Company, M. Baiget, P. Gallano

POSTER SESSION 3:
INCLUSION BODY MYOSITIS & HEREDITARY INCLUSION-BODY MYOPATHIES

S8

SM31. Resveratrol, a naturally occurring polyphenol found in red wine, increases Cathepsin D activity in endoplasmic reticulum stress (ERS)-provoked cultured human muscle fibers (CHMFs): relevance to treatment of sporadic inclusion body myositis (s-IBM)
A. Nogalska, C. D’Agostino, W.K. Engel, V. Askanas

SM32. The multifaceted role of chaperones in inflammatory myopathy: muscle fiber regeneration versus the cytotoxic action of inflammatory cells
J. De Bleecker, B. De Paepe, K. Creus, J. Weis

SM33. Assessment on muscle satellite cell population in inclusion body myositis
A. Kubota, H. Kowa, A. Iwata, J. Shimizu, S. Tsuji

SM34. Immunohistochemical studies on HSP47 and vessel related proteins in inclusion body myositis and muscular dystrophies
I. Higuchi, Y. Inamori, K. Higashi, E. Matsuura, A. Hashiguchi, T. Shiraishi, N. Hirata, K. Arimura

SM35. Myopathologic studies of an autopsy case with inclusion body myositis (IBM)

SM36. Expression of VCP in muscle fibers of experimental chloroquine-induced myopathy

SM37. Dysregulation of myogenic regulatory factors and myo-endothelial remodelling in sporadic inclusion body myositis

SM38. Prevalence of inclusion body myositis (IBM) in Japanese population

SM39. Magnetic Resonance Imaging of skeletal muscles in sporadic inclusion body myositis
F. Cox, M. Reijnierse, C. van Rijswijk, A. Wintzen, J. Verschuuren, U. Badrising

SM40. Can regulatory T cells be beneficial to sporadic inclusion body myositis patients?
Y. Allenbach, J. Wanschitz, M. Rosenzwajg, S. Hersen, D. Klatzmann, O. Benveniste

S16

SM41. Natural history of Distal Myopathy with Rimmed Vacuoles with long duration (DMRV)
H. Tomimitsu, M. Kobayashi, N. Sanjo, T. Hattori, M. Obara, T. Kanouchi, H. Mizusawa

SM42. Muscle atrophy in the DMRV/hIBM mouse model
S. Noguchi, M.C.V. Malicdan, Y.K. Hayashi, I. Nonaka, I. Nishino

SM43. Explicit distal myopathy phenotype caused by VCP-gene mutation in a Finnish family
S. Sandell, T. Suominen, J. Palmio, O. Raheem, P. Hackman, S. Huovinen, H. Haapasalo, B. Udd
SM44. Can Treg cells be beneficial to Inclusion Body Myositis muscle?  
Y. Allenbach, J. Wunschitz, M. Rosenzweig, C. Bloch-Queyrat, S. Herson,  
D. Klatzmann, O. Benveniste
SM45. Late-onset autosomal dominant limb girdle muscular dystrophy  
and Paget’s disease of bone unlinked to the VCP gene locus  
M. Kottlors, O. Moske-Eick, A. Huebner, S. Krause, K. Mueller, W. Kress, R. Schwarzwald,  
A. Bornemann, V. Haug, M. Heitzer, J. Kirchner
SM46. Role of ubiquitin-proteasome proteolysis in muscle fiber destruction  
in experimental chloroquine-induced myopathy  
T. Kumamoto, N. Kimura, T. Oniki, M. Nomura, K. Nakamura, Y. Abe, Y. Hazama,  
H. Ueyama
SM47. Novel valosin containing protein mutation in a Swiss family with hereditary  
inclusion body myopathy, Paget’s disease of the bone and dementia  
AK Peyer, J. Kinter, S. Frank, P. Fuhr, A. Fischmann, S. Kneifel, S. Thomann,  
P. Camaio, M. Sinnreich, S. Renaud

POSTER SESSION 4:  
RECENT ADVANCES IN GENETICS AND THERAPEUTICS  
OF MYOTONIC DYSTROPHIES  
Chairpersons: G. Meola, C. Thornton

S9

SM48. Frequency of DM2 and DM1 mutations in the Finnish population  
T. Suominen, L. Bachinski, L. Peltonen, R. Krahe, B. Udd
SM49. Management of respiratory impairment in Steinert Disease  
M.G. Di Gregorio, G. Fiorentino, A. Annunziata, A. Zaccaro, M. Scutifero, R. Cauteruccio,  
A. Palladino, M. Caputi, G. Nigro, L. Politano
SM50. Respiratory involvement in a large group of DM1 patients:  
is there any predictive test?  
L. Morandi, V. Patruno, A. Pincherle, P. Raimondi, S. Moretti, A. Dominese,  
P. Di Benedetto, F. Villani, R. Mantegazza
SM51. The efficacy of Bachmann’s bundle stimulation in myotonic  
dystrophy type 1 patients  
G. Nigro, V. Russo, A. Papa, L. Delli Paoli, F. Di Meo, A. Merola, F. Colimodio,  
A. Palladino, M. Scutifero, M.G. Russo, P. Golino, R. Calabro, L. Politano
SM52. Relationships among organ involvements in myotonic dystrophy  
P. Kaminsky, M. Pousset, L. Pruna, B. Chemuel, B. Brembilla-Perrot
SM53. Cysteine donor dietary intake is effective in reducing oxidative stress  
in myotonic dystrophy type I: a double-blind study with placebo  
L. Volpi, G. Ricci, L. Pasquali, M. Falorni, A. LoGerfo, C. Tramonti, V. Calsolaro,  
E. Caldarazzo Ienco, M. Franzini, A. Paolicchi, G. Siciliano
SM54. Altered pre-mRNA processing in myotonic dystrophy type 2 (DM2) myoblasts  
L.V. Renna, M. Malatesta, F. Perdona, R. Cardani, M. Giagnacovo, C. Pellicciari, G. Meola
SM55. FISH on muscle biopsy is a rapid and reliable method for diagnostic screening  
of patients with clinical suspicion of myotonic dystrophy type 2 (DM2)  
G. Silvestri, M. Santoro, A. Modoni, M. Masciullo, M.L.E. Bianchi, G. Tasca, E. Ricci
SM56. High prevalence of tumors in myotonic dystrophy  
P. Kaminsky, L. Pruna
SM57. Central nucleation, type 2 atrophy and ribonuclear inclusions:  
muscle histopathological markers of myotonic dystrophy type 2  
E. Bugiardini, R. Cardani, L.V. Renna, E. Mancinelli, V. Sansone, G. Meola
SM58. Body composition in patients with myotonic dystrophy type 1  
P. Kaminsky, J. Chatelin, V. Pascal, L. Pruna
SM59. Acute respiratory failure caused by pulmonary thromboembolism in a myotonic dystrophy patient
J.Y. Cho, O. Kwon, N.H.Kim

SM60. Distribution and severity of muscle changes in myotonic dystrophy type 2, compared to type 1. A muscle ultrasound study
A. Tieleman, A. Vinke, S. Pillen, H. van Dijk, N. van Alfen, B. van Engelen

SM61. Calcium homeostasis in Myotonic Dystrophy type 1
E. Loro, E. Gianazza, A. Malena, F. Rinaldi, V. Romeo, C. Angelini, A. Botta, M. Brini, M. Mongillo, L. Vergani

SM62. Molecular pathomechanisms of increased risk of statin-induced myopathy in myotonic dystrophy type 2 patients
M. Screen, P. Hackman, R. Laaksonen, M. Sirito, R. Krahe, B. Udd

SM63. Characteristics of cerebral involvement in adult onset myotonic dystrophies
N.H. Kim, J.Y. Cho, O. Kwon

SM64. Atypical correlation between CTG repeat size variation and clinical manifestation in brothers of myotonic dystrophy
J.H. Han, D.E. Kim

SM65. Neoplasia in Myotonic Dystrophy type 1 – analysis of a centre
M. Cardoso, T. Coelho

SM66. The DM-Scope myotonic dystrophies (DM1/DM2) database: a dual application system for clinical management and scientific research
G. Bassez, C. Guiraud-Dogan, D. Hamroun, C. Beroud, J. Puymirat

SM67. Does Bachmann Bundle’s pacing prevent atrial fibrillation in myotonic dystrophy type 1 patients? A twelve months follow-up study

SM68. Colocalization of DMPK and ZNF9 transcripts with MBNL1 protein in differentiated tissues from patients with myotonic dystrophy
Z. Lukas, I. Falk, M. Falk, R. Hrabalkova, J. Zaoralkova, Z. Lukas

SM69. Abnormalities of nerve conduction studies in myotonic dystrophy type 1 and type 2
M. Banach, M. Rakowicz, R. Rola, J. Antczak

POSTER SESSION 5A:
MUSCULAR DYSTROPHIES: STATE OF THE ART IN DIAGNOSIS
Chairpersons: F. Muntoni, M. Vainzof

S12

SM70. Improvement of survival in Duchenne Muscular Dystrophy. Retrospective analysis of 516 patients
L. Passamano, M.R. Cecio, V. Torre, A. Taglia, G. Nigro, L. Politano

SM71. Dystrophin gene mutations and their clinical correlates in 319 Italian patients affected with dystrophinopathy
F. Magri, A. Govoni, R. Del Bo, M.G. D’Angelo, S. Ghezzi, S. Tedeschi, S. Gandossini, A. Bordoni, S. Corti, V. Lucchini, M. Moggio, N. Bresolin, G. Comi

SM72. Identification of insulin resistance and obesity in Duchenne/Becker muscular dystrophy
M. Rodríguez-Cruz, A. Sánchez García, M. Bernabe García, M. López Alarcón, A.C. Velázquez Wong, R. Coral Vázquez, R.E. Escobar

SM73. CARE-NMD: Dissemination and implementation of standards of care for Duchenne Muscular Dystrophy in Europe (including Eastern Europe)
V. Karcagi, A. Heczegfalvi, M. Garami, P. Vondracek, A. Kaminska, A. Lusakowska, A. Kostera-Pruszczyk

SM74. Osteopontin in Duchenne muscular dystrophy
L. Piva, B.F. Gavassini, M. Fanin, L. Bello, G. Sorarà, M. Ermani, E.P. Hoffman, C. Angelini, E. Pegoraro

SM75. Geno-phenotyping of a de novo silent DMD duplication in a carrier and her unborn child
D. Diodato, S. Sampaolo, A. Torella, O. Farina, F. Del Vecchio Blanco, V. Nigro, G. Di Iorio

SM76. Respiratory muscle function in Muscular Dystrophies
M.G. D’Angelo, M. Romei, S. Gandossini, D. Colombo, E. Marchi, S. Bonato, G.P. Comi, A.C. Turconi, A. Lo Mauro, A. Pedotti, N. Bresolin, A. Aliverti

SM77. Intermittent glucocorticoid therapy influence on adrenal function, bone density and body composition in children with duchenne dystrophy: a sixty months follow up prospective study
D. Fintini, A. D’Amico, C. Brufani, G. Ubertini, M. Cappa, E. Bertini

SM78. Quantitative evaluation of fatty infiltration in muscle in DMD patients using MRI
B. Wokke, J. Milles, A. Webb, A. Aartsma-Rus, J. Verschuuren, H. Kan

SM79. The ubiquitin ligase neuralized 2 is overexpressed in Duchenne Muscular Dystrophy
E. Gazzerro, R. Pilu, F. Fruscione, A. Bonetto, S. Assereto, C. Minetti

SM80. « Chipping away » for deletion/duplication analysis in muscle disease genes
F. Leturcq, M. Cossee, I. Marey, A. Vasson, B. Comba, V. Commere, J. Lemarchand, Y. Saillour, J. Chelly

SM81. Histopathological and transcriptional characterization of inflammatory features in FSHD muscles
G. Tosca, M. Pescatori, M. Mirabella, M. Monforte, T. Cubeddu, E. Iannaccone, R. Frusciante, E. Ricci

SM82. Unexpected high frequency of FSHD alleles in the general population
I. Scionti, M. Zatz, P. Arashiro, F. Greco, R. Tupler

SM83. A genotype-phenotype correlation study for FSHD: which factors influence the onset and progression of the disease?

SM84. About the facioscapuloperoneal muscular dystrophy in the K. kindred once again. Re-examination in 1993 of the famous K. kindred described for the first time by Oransky (1927). Clinical and molecular genetic study (withdrawn)
V. Kazakov, A. Skorometz, D. Rudenko, V. Kolynin

SM85. Depressed sarcomeric function in patients with facioscapulohumeral muscular dystrophy
C. Ottenheijm, G. Padberg, G. Stienen, B. Van Engelen

SM86. Non uniform fatty infiltration distribution of muscle tissue in facioscapulohumeral dystrophy
B. van Engelen, B. Janssen, R. Arts, N. Voet, C. Nabuurs, G. Padberg, A. Heerschap

SM87. SNP-whole genome homozygosity mapping: a quick and powerful tool to achieve an accurate diagnosis in LGMD2 patients

SM88. Mutation analysis of Limb girdle muscular dystrophies in the Czech Republic
Fajkusova, Lenka; Stehlikova, Kristyna; Vondracek, Petr; Hermanova, Marketa

42
SM89. “Reverse-engineering“ gene network as the process to identify potential dysferlin interactions
M. Cacciottolo, V. Belcastro, S. Laval, K. Bushby, D. Di Bernardo, V. Nigro

SM90. Dysferlin interacts with alpha-tubulin in skeletal muscle
B. Azakir, S. Di Fulvio, B. Erne, C. Therrien, M. Sinnreich

SM91. Dysferlin C2A domain is important for association with MG53 (TRIM72)
C. Matsuda, K. Kameyama, H. Takeshima, I. Nishino, Y.K. Hayashi

POSTER SESSION 5B:
MUSCULAR DYSTROPHIES: STATE OF THE ART IN DIAGNOSIS
& RNA MODULATION FOR DUCHENNE MUSCULAR DYSTROPHY
Chairpersons: M. De Visser, N. Levy

S12

SM92. Struggle for life in lethal muscular diseases ultimate achievements
Y. Rideau, L. Politano, G. Fiorentino, V. Riccio, A. Hahn

SM93. Clinico-pathological profile of dysferlinopathy
M. Kanikannan, C. Sundaram; S.M. Uppin, J. Afshan, R. Mridula, LSR Krishna, R. Borgohain

SM94. Lumping dysferlinopathy phenotypes based on clinical findings and muscle imaging studies

SM95. Muscle MRI findings of dysferlinopathy in Chinese patients
J. Xi, J. Lu, W. Zhu, S. Luo, B. Yin

SM96. Anoctaminopathy: Non-dysferlin Miyoshi type distal myopathy caused by mutated ANOS/TMEM16E
S. Penttilä, O. Raheem, T. Suominen, M. Röyttä, P. Hackman, B. Udd

SM97. Mitochondrial dysfunction and ubiquitin-proteasome pathway abnormalities in limb-girdle muscular dystrophy type 2A

SM98. Differential expression of microRNAs in primary and secondary calpainopathies
M. Aguennouz, O. Musumeci, C. Rodolico, A. Garufi, E. Barca, G. Vita, A. Toscano

SM99. Myospryn interacts with M-band titin and calpain 3

SM100. Mutations in the Fukutin-Related Protein Gene (FKRP) Clinical, structural and molecular genetical aspects of Limb Girdle Muscular Dystrophy type 2I (LGMD2I) in 43 Norwegian patients
S. Lindal, E. Stensland, C. Jonsrud, O. Nilsen

SM101. Fukutin-related protein localizes on the endoplasmic reticulum and perinuclear region in muscle of dystrophic animal models

SM102. Influence of caveolin-3 point mutations on crucial signalling pathways in vitro and in vivo model of caveolinopathy
E. Brauers, P. Martinez, M. De Baets, J. Weis

SM103. Rippling muscle disease (RMD): an autosomal dominant spanish kindred with a novel missense mutation in the caveolin-3 gene
J. Muñoz-Blanco, A. Cabello, M. Vorgard
SM104. Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy
H. Yukiko, C. Matsuda, I. Nonaka, I. Nishino

SM105. Skeletal muscle magnetic resonance imaging in oculopharyngodistal myopathy
H. Durmus, M. Dursun, F. Deymeer, Y. Parman, H. Lochmüller, P. Serdaroglu-Oflazer

SM106. Distribution of extra-ocularpharyngeal weakness in OPMD
N. Witting, A. Mensah, D. Milea, J. Vissing

SM107. Actin binding domain mutation in filamin C associated with a distinct distal myopathy
N. Laing, R. Duff, V. Tay, G. Ravenscroft, C. McLean, P. Kennedy, K. Reardon, D. Williams, J. Landers, B. Udd, K. Nowak

SM108. An Italian case of hereditary myopathy with early respiratory failure (HMERR) not associated with the titin kinase domain R279W mutation
G. Tasca, M. Monforte, M. Mirabella, A. Broccolini, R. Renna, M. Sabatelli, P. A. Tonali, B. Udd, E. Ricci

SM109. A sarcolemmal wounding assay to study membrane resealing in human cultured myoblasts
B. Azakir, B. Erne, M. Sinnreich

SM110. Novel myosin heavy chain immunohistochemical double staining developed for the routine diagnostic separation of I, IIA and IIX fibers
O. Raheem, S. Huovinen, T. Suominen, H. Haapasalo, B. Udd

SM111. Proteome analysis of molecular changes in muscle tissue of TMD titinopathy
P. H. Jonson, J. Sarparanta, A. Vihola, P. Hackman, B. Udd

S17

SM112. Clinical characterization of Becker Muscular Dystrophy patients with a “skipped” mutation

SM113. Current progress with the systemic administration trial of AVI-4658, a novel Phosphorodiamidate Morpholino Oligomer (PMO) skipping dystrophin exon 51 in Duchenne muscular dystrophy (DMD)
S. Shrewsbury, S. Cirak, M. Guglieri, K. Bushby, F. Muntoni

SM114. VLA-4 is a severity-related biomarker and a target for therapy in Duchenne muscular dystrophy

Note: Posters numbered from SM01 to SM69 and from SM121 to SM167 will be discussed on Sunday; Posters numbered from SM70 to SM114 and from SM168 to SM224 will be discussed on Monday.

Monday – July 19th, 2010

h. 8.00 – 10.00  Poster viewing, coffee and rolls

h. 10.00 – 12.00  PLENARY LECTURES (Red and Blue Halls)
Chairs: Irena Hausmanowa-Petrusewicz (Poland) and Lewis P. Rowland (USA)

h. 10.00 – 10.30  PL5. The translational potential of immunomodulatory therapy in amyotrophic lateral sclerosis
Stanley Appel (USA)
h. 10.30 – 11.00  PL6. State of the art in muscle lipid diseases
Ichizo Nishino (Japan)

h. 11.00 – 11.30  PL7. State of the art in human Muscular Dystrophy.
Kate Bushby (UK)

h. 11.30 – 12.00  PL8. A rational approach to diagnosing and interpreting inherited neuropathies
Michael Shy (USA)

h. 12.15 – 14.00  Lunch (self-served buffet)

h. 14.00 – 15.00  MEET THE PROFESSOR – 2 CONCURRENT SESSIONS

MTP3 (Red Hall) Muscle Pathology

MTP4 (Blue Hall) Pediatric Neuromuscular Diseases
G. Serratrice and F. Muntoni (Moderators) – E. Bertini – C. Minetti

h. 15.10 – 17.00  SIMULTANEOUS SYMPOSIA (S8-S14)

S8 (Red Hall)  Inclusion-Body Myositis
Chairs: Valerie Askanas and Marinos Dalakas

Invited Speakers

S8.L1. Sporadic Inclusion Body Myositis: Clinical features and genetic predisposing factors
Frank Mastaglia

S8.L2. Defective muscle regeneration in s-IBM.
Massimiliano Mirabella

S8.L3. Putative pathogenic role of the endoplasmic reticulum stress in Sporadic-Inclusion Body Myositis (s-IBM)
Anna Nogalska

Platform Presentations

S8.PP1. p62-immunoreactive inclusions provide a better diagnostic marker of sporadic inclusion body myositis (s-IBM) patients’ muscle biopsies than TDP43 immunoreactivity
C. D’Agostino, A. Nogalska, W.K. Engel, V. Askanas

S8.PP2. Inhibition of the p97/VCP/Cdc48 ATPase promotes muscle growth and retards atrophy by inhibiting proteolysis via the ubiquitin-proteasome and the autophagic/lysosomal pathways
R. Piccirillo, A. Goldberg

S8.PP3. Immunosuppressive-treatment does not alter natural history of sporadic inclusion body myositis (sIBM): the Pitié-Salpêtrière/Oxford study
S9 (Hall E1) Recent advances in genetics and therapeutics of Myotonic Dystrophies
Chairs: Charles Thornton and Giovanni Meola

Invited Speakers

S9.L1. Persistence pays off: emerging phenotypes in mice that carry the human DM1 locus
Geneviève Gourdon

S9.L2. Regeneration defect and senescence in myotonic dystrophy muscle cells
Denis Furling

S9.L3. Clinical and pathogenic aspects of Myotonic Dystrophy Type 2
Giovanni Meola

S9.L4. Antisense approaches for myotonic dystrophy type 1
Jack Puymirat

Platform Presentations

S9.PP1. Differential miRNAs expression profiles in myotonic dystrophy type 1
R.Cardani, R. Perbellini, S.Greco, G. Sarra-Ferraris, G. Meola, F. Martelli

S9.PP2. Drug screening for Myotonic dystrophy type 1 (DM1)
J. Kinter, M. Sinnreich

S10 (Hall E2) Mitochondrial Encephalomyopathies
Chairs: Salvatore Di Mauro and Michio Hirano

Invited Speakers

S10.L1. Therapeutic strategies in mitochondrial encephalopathies
Michio Hirano

S10.L2. mtDNA gene defects in mitochondrial encephalomyopathies
Anders Oldfors

S10.L3. Nuclear gene defects in mitochondrial encephalomyopathies
Massimo Zeviani

Platform Presentations

S10.PP1. Genotype-phenotype correlations and muscle histopathology in patients with mutations in the TP53 gene

S10.PP2. MELAS and L-arginine therapy. Five years outcome on 8 MELAS patients

S11 (Hall F1) Extracellular Matrix Myopathies
Chairs: Luciano Merlini and Robert Griggs
Invited Speakers

S11.L1. Apoptosis and autophagy in human and murine COL6 myopathies
Paolo Bonaldo

S11.L2. Clinical aspects of extracellular matrix myopathies
Luciano Merlini

Patrizia Sabatelli

Platform Presentations

S11.PP1. Duchenne muscular dystrophy fibroblasts have a profibrotic phenotype and are more resistant to apoptosis
M. Mora, S. Zanotti, S. Gibertini

S11.PP2. Endomysial fibrosis in DMD: a marker of poor outcome associated with macrophage alternative activation
I. Desguerre, C. Christov, M. Mayer, R. Gherardi

S12 (Blue Hall) Muscular Dystrophies: state of the art in diagnosis (in honour of George Karpati)
Chairs: Valerie Cwik and Kate Bushby

Invited Speakers

S12.L1. Protein led diagnostic testing for the limb-girdle muscular dystrophies
Rita Barresi

S12.L2. High throughput molecular screening for muscular dystrophies
Nicolas Levy

S12.L3. How to make a molecular diagnosis of facio-scapulo-humeral dystrophy
Silvère van der Maarel

Platform Presentations

S12.PP1. Splice site strength and nonsense-associated exon skipping in the DMD gene
K. Flanigan, D. Dunn, J. Mendell, A. Pestronk, J. Florence, UDP Consortium, R. Weiss

G. Piluso, M. Dionisi, A. Torella, F. Del Vecchio Blanco, S. Aurino, V. Nigro

S13 (Hall F2) Animal models to define molecular mechanisms of ALS
Chairs: Stanley Appel and Gioacchino Tedeschi

Invited Speakers

S13.L1. Pathological events in fALS mice: from mitochondrial dysfunction to protein aggregation
Caterina Bendotti
S13.L2. Immunotherapy in models of ALS  
Jean Pierre Julien

Wim Robberecht

Platform Presentations

S13.PP1. Systemic transplantation of c-kit+ cells ameliorates the phenotype of a mouse model of amyotrophic lateral sclerosis  

S13.PP2. Corpus callosum DTI as a biomarker for amyotrophic lateral sclerosis  
N. Filippini, C. Mackay, G. Douaud, S. Knight, K. Talbot, M. Turner

S. Marcuzzo, P. Cavalcante, C. Cappelletti, I. Zucca, P. Bernasconi, R. Mantegazza

S14 (Hall F4)  
Nuclear envelope diseases  
Chairs: Gisèle Bonne and Alan E.H. Emery

Invited Speakers

S14.L1. Laminopathies affecting the striated muscles: from gene defects to possible pathophysiological mechanisms  
Gisèle Bonne

S14.L2. The congenital spectrum of laminopathies  
Luisa Politano

S14.L3. Loss of lamina-associated polypeptide 2 apha affects satellite cell differentiation and impairs heart function  
Roland Foisner

Platform Presentations

S14.PP1. Prelamin A-SUNs interplay in human muscle cells  
G. Lattanzi

S14.PP2. Characterization of Emd−/−/LmnaH222P/H222P mice: focus on the heart  

S14.PP3. Mutation of SYNE-1, encoding an essential component of the nuclear lamina, is responsible for autosomal recessive arthrogryposis  
R. Attali, N. Warwar, A. Israel, I. Gurt, E. McNally, M. Puckelwartz, B. Glick, Y. Nevo, Z. Ben-Neriah, J. Melki
POSTER PARALLEL SESSIONS (6 – 8)

POSTER SESSION 6A:
PATHOGENIC MECHANISMS OF INHERITED NEUROPATHIES
Chairpersons: M. Reilly, M. Shy

S2

SM121. Intrafamilial CMT1A variability among members of a numerous five
generation Polish family
M. Banach, A. Kochański, I. Hausmanowa-Petrusewicz

SM122. Charcot-Marie-Tooth disease type 2E: an Italian family with
GLU396LYS mutation
A. Mazzeo, L. Gentile, G.M. Fabrizi, T. Cavallaro, F. Taioli, S. Monaco, F. Granata,
A. Toscano, G. Vita

SM123. The clinical spectrum of X-linked Charcot-Marie-Tooth disease in childhood

SM124. Variable severity of early onset CMT2 with compound heterozygous
MFN2 mutations
M. Laura, J.P. Polke, D. Pareyson, F. Taroni, M. Milani, J. Blake, V.S. Gibbons,
C. Devile, M.G. Sweeney, M.B. Davis, M.M. Reilly

SM125. Characterisation of MFN2 gene mutations in Italian autosomal dominant
axonal CMT families
P. Nicolaou, E. Zamba-Papanicolaou, L. Politano, L.T. Middleton, G. Nigro,
K. Christodoulou

SM126. Variable phenotypes are associated with PMP22 missense mutations
M. Laura, M. Russo, J.P. Polke, M.B. Davis, J. Blake, S. Bradner, R. Hughes,
H. Houlden, M. P.T. Lunn, M.L. Reilly

SM127. Clinical variability of CMT 1 in a cohort of 15 patients of 6 different
albanian family pedigree
K. Altim, G. Vyshka, J. Kruja

SM128. A new French family with GDAP1-related autosomal dominant
Charcot-Marie-Tooth disease

SM129. The phenotype of the Gly94fsX222 PMP22 insertion
S. De Vries, F. Van Ruissem, C. Verhamme, W.F. Arts, H. Mauser, H. Kerkhoff,
B. Van Engelen, M. Lammens, M. De Visser, F. Baas, A. Van der Kooi

SM130. Coexistence of CMT1A and diabetes – A morphometric study
K.P. Lin, Y.C. Lee

SM131. Oral curcumin treatment of the R98C knock-in mouse model of CMT1B
A. Patzko, I. Katona, M.A. Saporta, A. Jani-Acsadi, L.M. Dillon, M. Shy

SM132. Neuregulin I type III overexpression does not rescue the phenotype
of the R98C knock-in mouse model of CMT1B
A. Patzko, X.Y. Wu, I. Katona, L.M. Dillon, J. Kamholz, M. Shy

SM133. Amino acid substrate selectivity alters desoxy sphingoid bases and
disease severity in HSAN1
F. Eichler, K. Garofalo, T. Hornemann, B. Schmidt, R. Brown

SM134. High frequency of SH3TC2 (KIAA1985) mutations in Czech HMSN
I patients
P. Lassuthova, R. Mazanec, S. Prasilova, P. Vondracek, D. Siskova, J. Haberlova,
P. Seeman

SM135. A new founder GDAP1 (Ala156Gly) mutation segregates with autosomal dominant
CMT4A disease
D. Kabzińska, J. Pilch, H. Strugalska-Cynowska, I. Hausmanowa-Petrusewicz,
A. Niemann, N. Huber, U. Suter, A. Kochański
SM136. Unfolded protein response (UPR) activation in a mouse model of early-onset CMT 1B
M. Saporta, Y. Bai, B. Shy, A. Patzko, M. Pennuto, C. Southwood, A. Gow, C. Ferri, M.L. Feltri, M. Crowther, D. Kirschner, L. Wrabetz, M. Shy

SM137. Added value of radial sensory nerve action potential (SNAP) in the CMT Neuropathy Score
M. Saporta, M. Laura, A. Saporta, C. Siskind, L. Miller, S. Feely, J. Blake, R. Lewis, M. Reilly, M. Shy

SM138. TRPV4 mutants in axonal disease localize appropriately and induce toxicity due to a gain of constitutive function
F. Fecto, Y. Shi, H.X. Deng, C. Klein, T. Siddique

SM139. Mutations in MPZ and PMP22 genes in hereditary motor sensory neuropathies
T.V.S. Mohan Vamsi, K. Meena, A. Jyothsna, S. Aparna, R. Renuka, M.S. Lavanya, M.D. Sadhanani

SM140. Mosaicism for GJB1 (Cx32) mutation causes milder Charcot-Marie-Tooth type X1 (CMTX1) phenotype in heterozygous man than in full carrier woman
P. Seeman, R. Mazanec, I. Sakmaryová, E. Koèárek, L. Baránková, M. Havlova

SM141. Charcot Marie Tooth Disease and Neuromyotonia
Y. Parman, E. Shugaiv, Z. Matur, M. Poyraz, P. Serdaroglu-Öflazer, F. Deymeer

SM142. The mutational spectrum of axonal Charcot-Marie-Tooth disease in Chinese population on Taiwan
Y.C. Lee, K.P. Lin

SM143. Clinical, neurophysiological and histopathological studies in IV drug abusers in East Delhi
V.N. Mishra, K. Bala, H.S. Malhotra, S. Kushwaha, V. Pardarsani, R. Kumar, N. Kumar

POSTER SESSION 6B:
PATHOGENIC MECHANISMS OF INHERITED NEUROPATHIES
Chairpersons: G. Di Iorio, S. Sampaolo

SM144. Pulse-wave velocity and nerve conduction study in diabetic patients
Y.B. Kim, B.C. Suh, P. Chung, H.S. Moon

SM145. Correlation between palmar bowing of the flexor retinaculum and serum lipid level in carpal tunnel syndrome patients: A preliminary study
J. In Soo, Y. Seung Hyeon

SM146. Neuromuscular electrical stimulation in management of low back pain: two controlled clinical trials in Ufa, Russia
L. Akhmadeeva, G. Rayanova, N. Setchenkova, B. Veytsman

SM147. Catamenial radiculopathy and plexopathy – Ectopic endometriosis
J. Vanderpol

SM148. Outcome of vitamin C supplementation on lead-induced apoptosis in adult rat hippocampus
M. Mehdi, F. Kermanian, I. Nourmohamadi

SM149. Lower extremity weakness and dysesthesias associated with zinc deficiency
S. Iyadurai, T. Bear, G. Lopate

SM150. Acute disturbance of axonal excitability after controlled alcohol intake
T. Prell, B. Winter, O.W. Witte, J. Grosskreutz

SM151. Botulism outbreak north in Iran: Five cases of a family
F. Abbasi, P. Vahdani, H.R. Behzad, M. Beshart, M. Haghighi, K. Aghazadeh Sarhangipour, S. Korooni
SM152. A case report – An unusual clinical presentation in a family with hereditary neuromuscular amyotropy - expansion of the phenotype
D. Lev, T. Sagie, M. Sadeh, E. Lysinsky-Silver, M. Ginsberg

SM153. Structural and functional changes in muscle fibers of patients with acquired spasticity
V. Kurushin, I. Ogneva, B. Shenkman

SM154. Brachial plexopathy complicating Epstein-Barr virus infection in an adult
D. Dodig, M. L. Ngo, D. Bailey, J. Karuviella, V. Bril

SM155. Acute tetraparesis in a pregnant woman as onset of an acute intermittent porphyria

SM156. Prevalence of tarsal tunnel syndrome in patients with fibromyalgia: comparison with normal controls

SM157. Acute facial diplegia and paraesthesias following Parvovirus B19 (HPV-B19) infection
A. Ariatti, F. Barbi, K. Funakoshi, E. Canali, S. Meletti, M. Meacci, M. Odaka, G. Galassi

SM158. Proximal ulnar nerve lesion with Martin-Gruber anastomosis masquerading ulnar neuropathy at the elbow
O. Kwon, N. H. Kim, J. Y. Cho

SM159. The role of provocative tests in diagnosis of cervical radiculopathy
M. Ghasemi, S. A. Mousavi, K. Golabchi, B. Asadi, F. Khovash

SM160. Quantitative analysis of somatic and autonomic small fibers in diabetic neuropathy
E. H. Sohn, A. Y. Lee, H. J. Kim, K. S. Song

SM161. Diabetic peripheral neuropathic pain in recent type 2 diabetes mellitus
S. Bursova, E. Vlkova-Moravcova, M. Nemec, J. Belobradkova, P. Dubovy, I. Klusakova, J. Bednarik

SM162. The influence of low intensity laser therapy on neuropathic pain and nerve sensory conduction velocities values in patients with diabetic neuropathy
Z. Peric

SM163. Altered nerve excitability properties in early diabetic neuropathy
J. S. Bae, S. H. Lee

SM164. Four novel peripheral myelin protein 22 gene (PMP22) mutations in Czech HNPP and Dejerine Sottas neuropathy patients
J. Haberlova, R. Mazanec, Z. Rychly, J. Sabova, J. Lisofova, P. Seeman

SM165. Pulsed high-dose dexamethasone versus standard prednisolone treatment for chronic inflammatory demyelinating polyradiculoneuropathy (PREDICT study): a double-blind, randomised, controlled trial.

SM166. Rituximab treatment in non-malignant inflammatory sensory polyganglionopathy
T. Kuntzer, A. Menetrey, J. Kleeberg

SM167. Syringomyelia-like phenotype of tangier disease: clinical, electrophysiological and genetic findings
A. Behin, T. Stojkovic, J. Zyss, C. Vial, A. Lacour, F. Bouhouri, A. Sassolas, C. A. Maurage, F. Couvert

POSTER SESSION 6C:
PATHOGENIC MECHANISMS OF INHERITED NEUROPATHIES & FRIEDREICH ATAXIA
Chairpersons: R. Baloh, I. Illa

SM168. Hereditary neuropathy with liability to pressure palsy
A. Q. Rana
SM169. Demyelination of sural nerve biopsies in Churg-Strauss syndrome
M. Kobayashi, H. Tomimitsu, T. Yokota, H. Mizusawa

SM170. Vascular involvement in a case of sarcoidosis with neuropathy and myopathy
C. Vital, P. Mercie, A. Vital

SM171. Marinesco Sjögren syndrome (MSS): Novel SIL1 mutations and description of a less severe phenotype

SM172. Axonal deformities in a mouse model of Hereditary Neuropathy with liability to Pressure Palsies (HNPP)
M. Saporta, I. Katona, Y. Bai, M. Shy, J. Li

SM173. Clinical and immunologic spectrum of anti-GQ1b antibody syndrome
Y.M. Lim, K.K. Kim

SM174. Readthrough acetylcholinesterase C-terminal peptide and PKCβII accumulation in the sciatic nerve following exposure to inflammatory medium

SM175. The first Serbian family with Familial Amyloidotic Polyneuropathy (FAP) (withdrawn)
I. Marjanovic, D. Lavrnic, I. Basta, A. Nikolic, Z. Stevic, V. Rakocevic, S. Peric, I. Boricic, S. Apostolski

SM176. Acquired amyloid neuropathy in a Portuguese patient after domino liver transplantation
I. Conceição, T. Evangelista, J. Castro, A.R. Silvestre, M. de Carvalho

SM177. C-Jun expression in human neuropathies: A pilot study

SM178. An atypical case of childhood CIDP
M. Ryan, A. Mohamed, V. Rodriguez-Casero

SM179. Nerve regeneration in a rare clinical context: Human T-lymphocyte virus (HTLV-1) associated cervical myelopathy and concurrent chronic inflammatory demyelinating polyneuropathy (CIDP)
V. Serban

SM180. Familial amyloidotic neuropathy TTR Val30Met a search for modifier genes

SM181. The signal transduction pathway of lead exposure induced β-amyloidogenesis and its neuronal damage in human neuroblastoma cell model (withdrawn)
S.S. Chen, C.T. Chen, T.J. Chen

SM182. Disruption of the Csn5/Jab1 gene in Schwann cell alters differentiation and survival causing a peripheral neuropathy

SM183. Pattern of onion bulb distribution may be predictive of acquired or of hereditary demyelinating polyneuropathy
J. Tracy, P. Dyck, C. Klein, J.N. Engelstad, J. Norell, P.J.B. Dyck

SM184. Guillain-Barre syndrome variant with hyperreflexia and antiganglioside antibodies
G. Patramani, C. Vogiatzi, S. Ralli, V. Gourgali, E. Dardiotis, G.M. Hadjigeorgiou, A. Papadimitriou

SM185. Very early electrodiagnostic findings in Guillain-Barré
M.A. Albertí, C. Casasnovas, A. Alentorn, M. Povedano, J.A. Martinez-Matos, J. Montero

SM186. The Glu89Lys gene mutation is characterized by early onset cardiac involvement and late neurological complication
F. Ochsner

SM187. Diagnostic validity of current perception threshold test in small fiber neuropathy
N.H. Kim, J.H. Min
SM188. Complement factor H and PMP22-related neuropathies
B. van Paassen, F. van Ruissen, E.J. Bradley, V. Timmerman, P. De Jonghe,
A.J. van der Kooi, F. Baas

S25

SM189. Cell models of Friedreich’s Ataxia (FA) and Amyotrophic
Lateral Sclerosis (ALS) derived from neural tissue of human olfactory mucosa,
to narrow down therapeutic agents
J.L. Muñoz-Blanco, I. Catalina, E. Scola-Plieg, M. Javier, F. Lim,
M. Rodríguez-Mahou, R. Jiménez-Bautista, R. Moure, J. Díaz-Nido

SM190. Results of a 6-month US phase 3 study evaluating efficacy and safety
of idebenone in children with Friedreich’s Ataxia
D.R. Lynch, S. Perlman, T. Meier

SM191. Abnormal brain iron accumulation in Ataxia with
Vitamin E Deficiency (AVED)
S. Airoud-Driss, L. Dellefave, T. Siddique

SM192. Huntington’s disease and ataxia symptoms in the same family:
searching a genetic modifier for cognitive impairment
M. Lazar, M. Haddad, M. Machado, R. Pavanello, F. Kok, C. Bueno, S. Amorim,
M. Martyn, E. Leão, N. Vieira, M.A. Alencar, M. Zatz

SM193 Screening for PRICKLE1 mutations in southern italian patients
with autosomal recessive progressive myoclonus epilepsy-ataxia syndrome
C. Criscuolo, M.F. de Leva, P. Sorrentino, R. Carbone, A. Guacci, G. De Michele, A. Filla

POSTER SESSION 7:
UPDATE ON LIPID MUSCLE DISEASES & UPDATE ON MUSCLE GLYCOGENOSIS
Chairpersons: C. Bruno, C. Navarro

S3

SM194. Fatty acid oxidation is impaired in patients with Neutral Lipid Storage
Disease with myopathy
M.C. Ørngreen, P. Laforêt, G. Andersen, N. Preisler, J. Vissing

SM195. Novel mutations and genotype-phenotype correlations in a large cohort
of muscular-type CPT-II deficient patients
D. Cassandrini, A. Anichini, M. Fanin, C. Minetti, C. Angelini, C. Bruno

SM196. Substrate reduction therapy (SRT) with Miglustat in a patient
with chronic GM2 gangliosidosis type Sandhoff: results of a three-years follow-up.
M. Masciullo, M. Santoro, A. Modoni, G. Guitton, E. Ricci, V. Rizzo,
PA. Tonali, G. Silvestri

SM197. Genetic variation and an increasingly personalised approach to medicine:
the Lysosomal Storage Disorder Registries
S. Prasad, E. James, F. Stewart

S15

SM198. Making diagnosis of Pompe disease at a presymptomatic stage:
to treat or not to treat?
J. Laloui, C. Wary, R.Y. Carlier, J.Y. Hogrel, C. Caillaud, M. Fardeau,
B. Eymard, P. Laforêt

SM199. Muscle phosphorylase kinase deficiency (Glycogenosis type VIII);
a neutral metabolic variant or a disease?
N. Preisler, M.C. Ørngreen, A. Echaniz-Laguna, P. Laforêt, E. Lonsdorfer-Wolf,
S. Doutreleau, B. Geny, S. DiMauro, J. Vissing

SM200. Role of the autophagic process in adult onset patients with Pompe disease
O. Musumeci, M. Aguennouz, T. Mongini, L. Palmucci, E. Barca, G. Vita, A. Toscano
SM201. Autophagy-related atrophy in different glycogen storage disease type II phenotypes: from a survival response to a pathogenetic mechanism
A.C. Nascimbeni, M. Fanin, E. Masiero, E. Tasca, C. Ferrati, M. Sandri, C. Angelini

SM202. An exploratory analysis of scoliosis in 182 children and adults with Pompe disease from the Pompe Registry
L. Merlini, L. Case, P. Kishnani, W. Muller-Felber, M. Roberts, A. van der Ploeg, S. Prasad

SM203. Quantitative metabolome profiling of biopsied muscle in the patients with glycogen storage diseases using capillary electrophoresis mass spectrometry
T. Fukuda, Y. Sugie, H. Sugie

SM204. Adult Pompe disease: bone mineral density before and after enzyme replacement therapy

SM205. A case of glycogen storage disease type III with a novel AGL mutation
Y.M. Lim, B.H. Kang, K.K. Kim

SM206. Alternative outcome measures in Pompe disease. Early changes after ERT
A. Dubrovsky, R. Peidro, F. Chloca, J. Corderi, M. Rugiero

M207. Debrancher deficiency, Glycogenosis type IIIa; a case of severe exercise intolerance and dynamic exercise-related symptoms
K.L. Madsen, N. Preisler, M.C. Ørngreen, G. Andersen, J. Vissing

SM208. Effects of the antioxidant N-acetyl-cysteine on the efficacy of enzyme replacement in Pompe disease fibroblasts
C. Porto, A. Luciani, M. Rosa, E. Balletta, C. Gagliardo, G. Andria, L. Maiuri, G. Parenti

POSTER SESSION 8:
ANIMAL MODELS TO DEFINE MOLECULAR MECHANISMS OF ALS & AMYOTROPHIC LATERAL SCLEROSIS: GENETICS, ENERGY HOMEOSTASIS AND FRONTO-TEMPORAL DEMENTIA.
Chairpersons: S. Appel, G. Tedeschi

S13

SM209. Angiogenesis in the spinal cord of transgenic rats with motor neuron degeneration
H. Warita, M. Aoki, H. Mizuno, N. Suzuki, Y. Ittoyama

SM210. Investigations on the pathogenesis of the VAPB P56S mutation in a familial form of ALS and detection of novel VAPB isoforms
T. Nachreiner, E. Brauers, A. Krüttgen, J. Weis

SM211. A novel antioxidant: Intravenous injection of platinum nanoparticles improve neurological findings in a rat model of familial amyotrophic lateral sclerosis (FALS1)
H. Yoshida

S18

SM212. Antisense oligonucleotide mediated exon skipping to down-regulate SOD1 expression via nonsense mediated decay: A novel antisense strategy in SOD1 associated Amyotrophic Lateral Sclerosis
M. Rajik, P. Kathirvel, W.C. Yee

SM213. SMN2 gene deletion in the patients with sporadic amyotrophic lateral sclerosis

SM214. TDP-43 pathology in sporadic ALS occurs in motor neurons lacking the RNA editing enzyme ADAR2
SM215. Satellite cells dysfunction may contributes to impaired skeletal muscle regeneration in sporadic amyotrophic lateral sclerosis
G. Cenacchi, A. Scaramozza, M. Brini, V. Papa, L. Tarantino, L. Badioli De Giorgi, E. Pegoraro, G. Soraru, C. Angelini

SM216. Lower extemity amyotrophic diplegia (LAD): Prevalence and pattern of weakness
M. Dimachkie, I.M. Muzyka, R.J. Barohn, J.S. Katz, C. Jackson, Y. Wang, A. McVey, A. Dick, M. Pasnoor, T. Mozaffar, X. Zhao, J. Kissel, J. Rosenfeld

SM217. MRI characteristics in 48 patients with Brachial Monomelic Amyotrophy (Hirayama disease)
N. Atchavaram, A. Gowda, G. Goel, R. Shivashankar

SM218. The usefulness of central motor conduction time for the assessment of upper motor neuron function using transcranial magnetic stimulation in early amyotrophic lateral sclerosis
J.Y. An, J.I. Seok, M.S. Park, B.J. Kim, S.G. Han

SM219. FALS with FUS mutation in Japan with early onset, rapid progress and basophilic inclusion

SM220. Upper Motor Neuron and SNP rs1541160 of KIFAP3 correlation
V. Orsetti, E. Pegoraro, C. D'Ascenzo, G. Querin, A. Palmieri, M. Volpe, V. Cima, C. Angelini, G. Soraru

SM221. Phenotypic presentation of I113T SOD1 ALS
T. Siddique, S. Donkervoort, N. Siddique, S. Ajroud-Driss

SM222. Variability in age of onset and ethnicity in the FUS/TLS mutation positive ALS patient population
T. Siddique, S. Donkervoort, N. Siddique, J. Yan

SM223. Developing an information system for the care and monitoring of patients with amyotrophic lateral sclerosis (ALS) in the region of Madrid (Spain): Population-based “Madrid ALS Register”

SM224. Immunohistochemical studies of TDP-43 in skin biopsies of patients with sporadic amyotrophic lateral sclerosis

Note: Posters numbered from SM01 to SM69 and from SM121 to SM167 will be discussed on Sunday; Posters numbered from SM70 to SM114 and from SM168 to SM224 will be discussed on Monday.

h. 18.30 – 19.00 Poster removal

Tuesday – July 20th, 2010

h. 8.00 – 10.00 Setting up Posters (TW), coffee and rolls

h. 10.00 – 12.00 PLENARY LECTURES (Red and Blue Halls)
Chairs: Michel Fardeau (France) and Frank Mastaglia (Australia)

h. 10.00 – 10.30 PL9. State of the art in human congenital muscular dystrophies.
Francesco Muntoni (UK)
PL10. Diagnosis and treatment of chronic immune mediated neuropathies.
Eduardo Nobile-Orazio (I)

Robert C. Griggs (USA)

PL12. Spinal muscular atrophy: animal and cellular models to elucidate pathogenesis and design therapeutic approaches
Judith Melki (France/Israel)

Lunch (self-served buffet)

MEET THE PROFESSOR – 2 CONCURRENT SESSIONS

MTP5 (Blue Hall) Veterinary Myology
J. Mc Howell (Moderator) – S. Blot – S. Papparella – D. Shelton – S. Valberg

MTP6 (Red Hall) Immuno-mediated disorders and Immuno-suppression

SIMULTANEOUS SYMPOSIA (S15-S21)

S15 (Hall E1) Update on muscle glycogenosis
Chairs: Antonio Toscano and Corrado Angelini

Invited Speakers

S15.L1. Update on therapeutic approach
Bruno Bembi

S15.L2. New insights of pathophysiology
Carmen Navarro

S15.L3. Typical and atypical phenotypes
Antonio Toscano

Platform Presentations

S15.PP1. The Italian Group on late-onset Glycogenosis type II: long term follow-up of 80 patients on Enzyme Replacement Therapy
C. Semplicini, T. Mongini, E. Pegoraro, M. Filosto, G. Marrosu, R. Piras, E. Sette,
M.A. Donati, S. Gasperini, G. Vita, G. Comi, M. Moggio, V. Lucchini, L. Morandi,
M. Rigoldi, R. Parini, G. Di Iorio, D. Diodato, M. Scarpa, G. Sciarabba, G. Crescimanno,
S. Ravaglia, G. Siciliano, L. Volpi, S. Servidei, R. di Giacopo, F. Giannini, L. Vercelli,
B. Bembi, P. Tonin, A. Toscano, C. Angelini

S15.PP2. The Angiotensin Converting Enzyme (ACE) Insertion/Deletion (I/D) polymorphism modifies the clinical outcome in Type II Glycogenosis (GSDII)
S. Ravaglia, P. De Filippi, B. Bembi, L. Piccolo, P. Bini, A. Moglia, A. Dardis,
G. Greco, G. Ciana, F. Canevari, C. Danesino
S15.PP3. Characterization of the regenerative potential of skeletal muscle in Pompe diseases (PD) mouse model
M. Cardone, B. Rossi, V. Saccone, A. Tarallo, G. Andria, G. Parenti

S16 (Hall E2)  
Hereditary inclusion-body myopathies
*Chairs: Stella Mitrani-Rosenbaum and Massimiliano Mirabella*

**Invited Speakers**

S16.L1. GNE-opathy: from bedside features to potential therapy
Zohar Argov

S16.L2. Experimental approaches in elucidating pathogenesis of h-IBM due to GNE mutations
Aldobrando Broccolini

S16.L3. h-IBM due to VCP mutation: clinical features, pathogenesis and experimental models
Virginia Kimonis

**Platform Presentations**

S16.PP1. Cell stress mechanisms in hereditary inclusion body myopathy (hIBM)
C. Fischer, I. Nishino, J. Schmidt

S16.PP2. Direct role of GNE in the early development of cardiac and skeletal muscle
S. Mitrani-Rosenbaum, I. Milman

S16.PP3. Sialic acid metabolites prevent the myopathic phenotype in the DMRV/hIBM mouse model

S17 (Red Hall)  
RNA modulation for Duchenne Muscular Dystrophy
*Chairs: Annemieke Aartsma-Rus and Eric Hoffman*

**Invited Speakers**

S17.L1. Efficacy and safety of ataluren (PTC124®) in nonsense mutation Duchenne/Becker muscular dystrophy (nmDBMD)
Leone Atkinson

S17.L2. Learning from early trials of antisense oligonucleotide therapy for DMD
Annemieke Aartsma-Rus

S17.L3. Clinical development of morpholinos for exon skipping in DMD
Steve Wilton
Platform Presentations

S17.PP1. Prednisolone treatment does not influence antisense-mediated exon skipping in DMD
I. Verhaart, H. Heemskerk, M. van Putten, S. de Kimpe, J. van Deutekom, GJ van Ommen, A. Aartsma-Rus

S17.PP2. Results of a Phase 2b study of ataluren (PTC124®) in nonsense mutation dystrophinopathy (Duchenne/Becker muscular dystrophy; nmDBMD)
K. Bushby, B. Wong, R. Finkel, K. Flanigan, C. McDonald, T. Voit, G. Spinella, V. Ćwik, A. Reha, G. Elfring, L. Miller, L. Atkinson

S18 (Hall F1) Amyotrophic Lateral Sclerosis: Genetics, energy homeostasis and fronto-temporal dementia.
Chairs: Orla Hardiman and Richard Barohn

Invited Speakers

S18.L1. Genome-wide SNP associations in sporadic ALS
Orla Hardiman

S18.L2. Insights into molecular basis of ALS-MND: from gene expression profiling
Pamela J. Shaw

S18.L3. Loss of stearoyl coenzymeA desaturase in skeletal muscle is an early event in ALS
Jean-Philippe Loeffler

S18.L4. The frontotemporal syndromes of Amyotrophic Lateral Sclerosis (ALS)
Michael J. Strong

Platform Presentations

S18.PP1. Mitochondrial respiratory chain dysfunction in muscle from patients with amyotrophic lateral sclerosis

J. Kirby, Ke. Ning, L. Ferraiuolo, P. Heath, A. Ismail, C. Valori, L. Cox, B. Sharrack, S. Wharton, P. Ince, P. Shaw, M. Azzouz

S19 (Hall F2) Mitochondrial Medicine
Chairs: Massimo Zeviani and Claude Desnuelle

Invited Speakers

Giulia D’Amati

S19.L2. Clinical details of Oculopharyngodistal myopathy
Piraye Serdaroglu

58
Claude Desnuelle

Platform Presentations

S19.PP1. Nuclear mitochondrial gene mutations in patients with chronic progressive external ophthalmoplegia and multiple mtDNA deletions  
J. Schaefer, D. Leupold, K. Witte, S. Jackson

T.D. Jeppesen, J. Vissing, J. Gonzales-Alonso

S19.PP3. Estrogens ameliorate mitochondrial dysfunction in Leber’s hereditary optic neuropathy  
C. Giordano, M. Montopoli, E. Perli, M. Orlandi, M. Fantin, A. Martinuzzi, A. Ghelli, L. Caparotta, F. Ross-Cisneros, A. Sadun, G. d’Amati, V. Carelli

S20 (Blue Hall) Gene therapy in muscular dystrophies  
Chairs: Francesco Muntoni and Serge Braun

Invited Speakers

S20.L1. Recombinant AAV-mediated gene transfer to the skeletal muscle of nonhuman primates: immunotoxicological issues and strategies to induce transgene immune tolerance  
Oumeya Adjali

S20.L2. Gene therapy of skeletal muscle diseases. Hopes and hipes  
Serge Braun

S20.L3. Sustained alpha-sarcoglycan gene expression for six months in LGMD2D following gene transfer  
Jerry Mendell

Platform Presentations

S20.PP1. A genetic therapy for dominant muscle disorders targeting RNA  
J. Wei, E. Stepiak, J.R. Chamberlain

S20.PP2. A vector-based approach to understand FSHD pathogenesis and develop potential RNAi therapies  
S. Harper, L. Wallace

S20.PP3. Phase I clinical study of AAV1-α-sarcoglycan gene therapy for limb girdle muscular dystrophy type 2C  
S21 (Hall F4)  
**Congenital Myopathies**  
*Chairs: Nigel Laing and Susan Iannaccone*

**Invited Speakers**

S21.L1. **What’s new in congenital myopathies**  
*Kathryn North*

S21.L2. **MRI differentiation of congenital myopathies**  
*Dirk Fischer*

S21.L3. **Molecular basis and pathological mechanisms of myotubular and centronuclear myopathies**  
*Jocelyne Laporte*

**Platform Presentations**

S21.PP1. **Centronuclear myopathies: the morphological particularities of genetically identified forms**  

S21.PP2. **New variant of myofibrillar myopathy with CNS involvement**  

S21.PP3. **Five new cases of congenital myopathy with hexagonally cross-linked crystalloid inclusions**  

**h. 17.00 – 18.30**  
**POSTER PARALLEL SESSIONS (9 – 13)**

**POSTER SESSION 9:**  
**NOVEL THERAPEUTIC TARGETS AT THE NEUROMUSCULAR JUNCTION & MYASTHENIC SYNDROMES**  
*Chairpersons: A. Evoli, L. Middleton*

S4  
**TW01. Lambert-Eaton myasthenic syndrome (LEMS) revealing intravenous multiple uterine leiomyomas: causal or incidental?**  
*G. Galassi, A. Ariatti, V. Agnoletto, E. Canali, F. Rivasi*

**TW02. Generalized weakness after focal injection of botulin toxin type A (BTA)**  
*G. Galassi, C. Orlandi, G. Albertini, A. Ariatti, F. Valzania, A. Barbieri*

**TW03. Clinical and demographical findings of MuSK myasthenia gravis in Argentina**  
*M. Rugiero, M. Bettini, V. Saluto, H. Gomez, L. Pirra, A. Dubrovsky, J. Politei, C. Mazia*

S22  
**TW04. Thymolipoma associated with myasthenia gravis**  

**TW05. Autoimmunity – myasthenia gravis, Graves’ disease and vitiligo: a case report**  
*V. Serban*

**TW06. The new cell-based assays are useful in the diagnosis of bulbar myasthenia**  
*M.E. Farrugia, I. Leite, R. Petty, J. Overell, A. Mallik, A. Vincent*
TW07. Study of clinical profile and long term outcome of myasthenia gravis in a tertiary care centre in western India (withdrawn)
N. Jain

TW08. Myasthenia Gravis (MG) associated with etanercept therapy (TNFα): causal or incidental?
A. Ariatti, L. Codeluppi, S. Meletti, G. Bigliardi, G. Galassi

TW09. Immune-mediated rippling muscle disease with Myasthenia Gravis: a report of three patients and long-term follow up
M. Rugiero, M. Bettini, E. Fulgenzi, A. Figueredo, V. Salutto, C. Mazia

TW10. Small cell lung cancer in the Lambert-Eaton myasthenic syndrome, creation and validation of a prediction model

TW11. HLA-DR3 and HLA-B8 are confined to early-onset non-tumour Lambert-Eaton myasthenic syndrome, similar to autoimmune myasthenia gravis

TW12. Marked clinical and electrophysiological response to fluoxetine treatment in congenital slow channel myasthenic syndrome
A.K. Peyer, A. Abicht, M. Sinnreich, D. Fischer

TW13. Autoantibodies in “seronegative” myasthenia gravis

TW14. Novel HLA class I and II associations in myasthenia gravis

TW15. Some clinical data of 75 myasthenia gravis patients
O. Sinanovic

TW16. Distal asymmetric weakness as a manifestation of myasthenia gravis
S. Ralli, P.M. Kountra, V. Tsimourtou, A. Markou, E. Dardiotis, G.M. Hadjigeorgiou, A. Papadimitriou

TW17. Antibodies against main immunogenic region (MIR) of the nicotinic acetylcholine receptor and ocular Myasthenia Gravis

TW18. Clinical and serological features of late-onset myasthenia gravis. A UK multicentre retrospective cohort study

TW19. MuSK-antibody positive Myasthenia Gravis and associated autoimmune diseases

TW20. Myasthenia Gravis followed by neuromyelitis optica spectrum disorders. Description of 8 Caucasian patients

TW21. Single-fiber electromyography (SFEMG) study in normal subjects & in patients with myasthenia gravis
A. Musa, M. Elzubair, R. Badi, A. Ahmed

TW22. Pharmacokinetics and tissue distribution of 3,4-diaminopyridine
K. Komai, R. Matsushita, Y. Kondo, N. Ishida, C. Ishida

TW23. The use of exon trapping to characterise DOK7 mutations
W.W. Liu, J. Cossins, D. Beeson
TW24. Active content variability of compounded 3,4-Diaminopyridine in solid oral dosage forms
D. Green, A.C. Jones, K. Brain

TW25. The epidemiology of congenital myasthenic syndromes in Northern Ireland
A. Carr, E. Healy, B. Herron, C. Cardwell, P. McCarron, D. O'Reilly, J. McConville

TW26. Significance of fine structures in motor end-plate in mice with a collagenous sub-unit called Q (ColQ)-deficient mutant mice

TW27. Expression of toll-like receptors 7 and 9 is increased in Epstein-Barr virus-positive thymus of myasthenia gravis patients
P. Cavalcante, C. Cappelletti, M. Barberis, F. Baggi, C. Antozzi, L. Maggi, S. Berrih-Aknin, R. Mantegazza, P. Bernasconi

TW28. Anti-MuSK positive patients indistinguishable from the anti-AChR positive ones regarding response to immunosuppressives
O. Gunog-Tuncer, V. Yilmaz, Y. Parman, P. Serdaroglu, G. Saruhan-Direskeneli, E. Deveer

TW29. Myasthenia gravis – an atypical presentation with dropped head syndrome and cognitive impairment

TW30. Lambert-Eaton myasthenic syndrome: Clinical and electrophysiological characteristics and response to treatment in 15 Iranian patients
S. Nafissi, R.H.B. Naeni, M.H. Haririan, A. Soltanzadeh, H. Sikaroodi

POSTER SESSION 10: VETERINARY MYOLOGY & ANIMAL MODELS FOR NEUROMUSCULAR DISEASES
Chairpersons: D. Shelton, M. Zatz

S7

TW31. Laminin alpha 2 deficiency in two Great Dane dogs: clinical, histological and molecular insights
F. Trapani, O. Paciello, S. Aurino, Stefania; L. Meomartino, G.D. Shelton, M.P. Pasolini

TW32. Histopathological changes in dog’s skeletal muscle due to aging
T. Pagano, F. Trapani, A. Costagliola, V. Iovane, S. Papparella, O. Paciello

TW33. Aging effects on skeletal muscle in cow
A. Costagliola, F. Trapani, T. Pagano, V. Iovane, S. Papparella, O. Paciello

S26

TW34. A transgenic mouse model of severe skeletal muscle actin disease

TW35. Mitochondrial ultrastructure in Golden Retriever Muscular Dystrophy (GRMD) dogs
C. Caetano, N. Vieira, T. Andrade, V. Silva, M. Zatz

TW36. Residual laminin-binding activity and enhanced dystroglycan glycosylation by LARGE in novel model mice to dystroglycanopathy
T. Toda, M. Kanagawa, A. Nishimoto, T. Chiyonobu, Y. Miyagoe-Suzuki, S. Takeda, T. Endo, K. Campbell, K. Kobayashi

TW37. Activin receptor type IIB inhibition improves strength and function of dystrophic muscle
J. Lachey, A. Pullen, A. Koncarevic, J. Seehra

TW38. Troponin T aberrant splicing explains muscle weakness in FSHD
R. Tupley, V. Sancisi, E. Germinario, A. Esposito, E. Morini, S. Peron, G. Tomelleri, D. Danieli
TW39. Mechano Growth Factor promotes cell proliferation in gastrocnemius muscle during recovery from chronic alcoholic myopathy in rats
O.V. Turtikova, E.A. Lysenko, V.P. Khrotchenkov, S.V. Benevolensky, V.O. Popov, B.S. Shenkman

TW40. Deposition of the Inner Limiting Membrane in the eye of a mouse model for Muscle Eye Brain disease
C. Whitmore, M. Ackroyd, I. Mavrommatis, S.C. Brown

TW41. Pre-clinical evaluation of the effects of a combined treatment with alpha-methyl-prednisolone and taurine in dystrophic mdx mouse
A. Cozzoli, J.F. Rolland, R.F. Capogrosso, V. Sbendorio, V. Longo, B. Nico, S. Simonetti, A. De Luca

TW42. Muscle pathology seen in older mixed inbred FVB; B6 Gne mouse model of hIBM

TW43. Functional and pathological comparison of mouse models for Duchenne Muscular Dystrophy
M. van Putten, D. Kumar, G.J. van Ommen, P.B. ’t Hoen, A.Aartsma-Rus

TW44. The dystrophin-glycoprotein complex plays a crucial role in myofiber regeneration
P. Konieczny, R. Ng, L. Judge, J.S. Chamberlain

TW45. Two pools of inorganic phosphate in canine model of DMD evidenced by phosphorus NMR spectroscopy
J.L. Thibaud, C. Wary, T. Naudet, S. Duteil, A. Monnet, S. Blot, P. Carlier

TW46. Resident macrophages crucially control muscle inflammatory reactions
M. Brigitte, F. Chretien, R. Gherardi

TW47. Muscle fibre profile number changes with age in mdx mice
R. Terry, N. Hasan, D. Wells

TW48. Role of oxidative stress in sarcolemmal damage and cytotoxicity: ex vivo and in vitro pharmacological studies on normal and dystrophic mdx mouse muscles and C2C12 cells
V. Sbendorio, R.F. Capogrosso, A. Cozzoli, A. De Luca

TW49. The mouse alpha-dystrobrevin gene is atypical; should we pursue non-murid rodent models of Duchenne muscular dystrophy?
R. Roberts, S. Boehm, L.L. Zhuo, R. Sewduth, P. Constantinou

TW50. From dysferlin deficiency to dysferlinopathy: the janus-faced role of exercise
F.J. Authier, O. Biondi, A. Marchand, A. Temchenko, F. Chretien, G. Bassez, N. Bourg, R. Gherardi, I. Richard

TW51. Tamoxifen is more potent than raloxifene to ameliorate skeletal muscle function of adult mdxscv dystrophic mice
O. Dorchies, J. Patte-Reutenauer, O. Patthey-Vuadens, U. Ruegg

TW52. Green tea polyphenols enhance motor performance and normalize calcium influx in a mouse model for Duchenne muscular dystrophy
O. Dorchies, C. Gallo, J. Patte-Reutenauer, E. Gayi, U. Ruegg

POSTER SESSION 11: EXTRACELLULAR MATRIX MYOPATHIES & MITOCHONDRIAL MEDICINE & NEW THERAPIES IN MUSCLE CHANNELLOPATHIES
Chairpersons: G. Lattanzi, A. Ferlini

S11

TW53. Characterization of Large transgenic mice -Overexpression of Large and functional up-regulation of α-dystroglycan in vivo
TW54. A reduction in the expression of fukutin related protein leads to the altered deposition of multiple laminin alpha chains in a mouse model for Muscle Eye Brain Disease
M. Ackroyd, C. Whitmore, S. Prior, U. Mayer, F. Muntoni, S. Brown

TW55. Expression of the Collagen VI α5 and α6 chains in human skin: implications for the pathogenesis of Collagen VI-related disorders
R. Curci, P. Sabatelli, S. Gara, P. Grumati, A. Urciuolo, F. Gualandi, S. Squarzoni, A. Zamparelli, E. Martoni, L. Merlini, M. Paulsson, P. Bonaldo, R. Wagener

TW56. Molecular and histopathological characterization of fibrosis in muscular dystrophies
S. Zanotti, E. Bellaﬁore, C. Di Blasi, S. Saredi, E. Mottarelli, S. Gibertini, R. Mantegazza, L. Morandi, M. Mora

TW57. Remodeling of the myotendinous junction during muscle regeneration: a cross-species approach through expression proﬁling
K. Gorni, Z. Po, L. Pasquali, E. Hoffman

TW58. Gene expression proﬁling of ﬁbroblasts to detect biomarkers for clinical variants of MND
R. Rohini; L.L. Ponger, S. Kramer, P. Heath, H. Hollinger, R. Hibberd, C. McDermott, J. Kirby, P. Shaw

TW59. TARDBP mutations, amyotrophic lateral sclerosis and alternative splicing in human ﬁbroblasts
R. Highley, J. Kirby, J. Jansweijer, P. Heath, P. Ince, P. Shaw

TW60. Blood macrophages as surrogate diagnostics in Collagen VI-related myopathies: implications for biomarker discovery
F. Gualandi, P. Sabatelli, R. Curci, E. Martoni, L. Merlini, A. Ferlini

S19

TW61. Clinical and pathological features of myopathy associated with antimitochondrial antibodies
M. Hashimoto, H. Kowa, A. Iwata, S. Tsuji, J. Shimizu

TW62. Mitochondrial disorders in a child with congenital fiber type disproportion
V. Sukhorukov, D. Kharlamov, D. Vlodavets, E. Belousova

TW63. Assessment of respiratory chain and antioxidant enzymes in skeletal muscle homogenates
M. Spinazzi, C. Angelini

TW64. A novel mitochondrial DNA mutation in the tRNA gene in a 12-year old boy with exercise intolerance
M. Meznaric, P. Gradisnik, B. Garavaglia, E. Lamantea, C. Lamperti, M. Zeviani

TW65. Oxidative stress biomarkers in mitochondrial myopathies, basally and after cysteine donor supplementation

TW66. Oxidative stress by monoamine oxidases is causally involved in myoﬁber damage in muscular dystrophy

TW67. Phenotypic expression of homoplasmic mutations in mitochondrial tRNA genes: role of aminoacyl-tRNA synthetases

S28

TW68. Differences in molecular composition, biophysical properties and drug responses of BK channels of slow- and fast-twitch skeletal muscle ﬁbers
M.M. Dinardo, A. Mele, G.M. Camerino, G. Cannone, R. Latorre, D. Conte Camerino, D. Tricarico
TW69. New mutations in the SCN4A gene associated with distinctive phenotypes
G. Silvestri, A. Modoni, S. Pagliarani, M. Lo Monaco, S. Lucchiari, A. D’Amico,
G.P. Comi, P.A. Tonali

TW70. A new mutation in SCN4A drastically alters phenotype
S. Portaro, E. Matthews, S.V. Tan, R. Sud, M.B. Davis, M.G. Hanna

TW71. The temporal expression pattern of TRPV2 involved in the myogenic differentiation of skeletal myoblast
Y. Katanosaka, S. Mohri, K. Naruse

TW72. Chronic non-paroxysmal neuropathic pain – Novel phenotype of mutation in the sodium channel SCN9A gene
R. Dabby, M. Sadeh, R. Gilad, Y. Lampl, S. Cohen, S. Inbar, E. Leshinsky-Silver

TW73. Hypokalemia with dropped hands or head: A myopathic or a neuropathic process? (withdrawn)
M.H. Chang

POSTER SESSION 12:
MITOCHONDRIAL ENCEPHALOMYOPATHIES
Chairpersons: M. Hirano, A. Oldfors

S10

TW74. Mitochondrial encephalomyopathy with sideroblastic anemia in two sibling patients: second report focusing on complex I deficiency
A. Hashiguchi, Y. Inamori, I. Higuchi, K. Higashi, H. Takakshima, K. Arimura

TW75. Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ10 deficiency
M. Schuelke, L. Salviati, R. Carrozzo, F. Santorelli, S. Rahman, M. Tazir, M. Koenig,
S. DiMauro, M. Hirano

TW76. Treatment of CoQ10 deficient fibroblasts with ubiquinone, analogs, and vitamin C: time- and compound-dependent effects on bioenergetic and oxidative stress status
R. Carrozzo, F. Santorelli, S. DiMauro, M. Hirano

TW77. Mutations in TMEM70, a gene coding for an assembly factor of complex V, can cause a severe encephalocardiomyopathy in the neonatal period as well as mild non-progressive encephalopathy in adolescence
R. Van Coster, J. Smet, W. Lissens, B. De Paepe, S. Seneca

TW79. A novel splice site mutation of the POLG gene in two unrelated adults with ophthalmoparesis and myopathy
M. Milong, T. Liewluck, J. Wang, J.A. Leavitt, L.J. Wong

TW80. Clinical experience with L-arginine treatment in MELAS syndrome; good response in young but not in adult-onset patients
A. Ishii, A. Shioya, Ai Hosaka, N. Ohkoshi, K. Nakamagoe, A. Tamaoka

TW81. Oxidative stress and apoptosis are extensive in Mitochondrial Encephalomyopathies and correlate with cytochrome c oxidase deficient fibers
T. Kyriakides, R. Charalambous, M. Feldman, A. Hadjisavvas, K. Kyriakou, L. Koutsou,
K. Christodoulou, I. Vonta, T. Dardiotis, G. Hadjigeorgiou, A. Papademitriou,
A. Kladi, P. Manda
TW82. Mitochondrial involvement in patients with mental retardation  

TW83. Incidental mitochondrial myopathy: five isolated cases  
V. Lucchini, V. Crucignola, L. Napoli, D. Ronchi, M.G.D’Angelo, A. Bordoni, G. Fagioli, L. Peverelli, N. Bresolin, G. Comi, M. Moggio, M. Sciaccio

TW84. MERRF mutation A8344G in a four-generation family without CNS involvement: clinical and molecular characterization  
V. Crucignola, L. Peverelli, V. Lucchini, A. Bordoni, P. Ciscato, V. Civelli, D. Spagnoli, N. Bresolin, G. Comi, M. Moggio, M. Sciaccio

TW85. Monozygotic twins with MERRF syndrome: clinical and genetic features  
M. Yger, C. Jardel, I. Lemiere, M. Guilleron, B. Eymard, P. Laforet

TW86. Maternally inherited limb-girdle myopathy with neck extensor and cardiopulmonary involvement resulting from a mitochondrial tRNA3302 point mutation  
T. Mozaffar, A. Wang, M. Simon, D. Wallace

TW87. Novel POLG mutation causing distal myopathy and cachexia  

TW88. Screening of the whole mtDNA mutations in 22 Chinese patients with MELAS  
Jie, Lin; Chongbo, Zhao; Jin, Zhang; Jianyin, Xi; Huijun, Wang; Wenhua, Zhu; Duan, Ma; Jiahong, Lu

TW89. Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G>A mitochondrial DNA mutation  
D. Ronchi, S. Orcesi, A. Bordoni, F. Fortunato, M. Moggio, A. Berardinelli, P. Veggio, G.P. Comi

TW90. Partial duplication of mtDNA-tRNAPhe in a patient with late-onset mitochondrial myopathy  
C. Lamperti, P. Arzuffi, M. Lombradi, M. Zeviani

TW91. New mutation in COI gene associated with CNS involvement and amenorrhoea  
C. Lamperti, E. Lamantea, R. Rizzi, C. Mariotti, F. Carrara, M. Zeviani

TW92. Analysis of stroke-like events in MELAS (Mitochondrial encephalomyopathy, Lactic Acidosis, and Stroke-like episodes): personal experience and review of the literature  
S. Servidei, R. Di Giacopo, A. Cianfoni, M. Catteruccia, D. Sauchelli, C. Cuccagna, G. Miceli

TW93. Mitochondrial Cardiomyopathy: new SCO2 gene mutations in a Brazilian patient  

TW94. Genotype-phenotype correlations of gastrointestinal dysmotility in mitochondrial encephalomyopathies  
C. La Morgia, R. De Giorgio, V. Stanghellini, G. Cenacchi, R. Rinaldi, L. Badiali De Giorgio, R. Cogliandro, A. Antonucci, L. Iommarini, P. Montagna, R. Liguori, V. Careelli, M.L. Valentino

TW95. Enteropathic arthritis in mitochondrial neurogastrointestinal encephalomyopathy  
TW96. A novel mutation in the anticodon of the mitochondrial tRNA
Arg in a patient with mitochondrial myopathy


TW97. Different clinical phenotypes associated with three new mutations in the cytochrome b gene

M.L. Valentino, L. Iommarini, L. Caporali, M. Pala, R. Liguori, P. Barboni,
M. Carbonelli, R. Carroccia, A. Maresca, F. Pizza, C. La Morgia, P. Avoni, A. Baruzzi,
P. Montagna, A. Achilli, A. Torroni, V. Carelli

POSTER SESSION 13:
BASIC MECHANISMS IN SPINAL MUSCULAR ATROPHY & SPINAL MUSCULAR ATROPHY: RECENT ADVANCES IN CLINICAL TRIALS & MISCELLANEOUS
Chairpersons: E. Bertini, E. Tizzano

S5

TW98. The study of the c.859G>C variant in chronic SMA patients confirms that is a milder SMN2 allele and predicts a direct correlation of SMN levels with phenotype

R. Martínez-Hernández, S. Bernal, L. Alias, E. Also-Rallo, M.J. Barceló, M. Baiget,
P. Fuentes-Prior, E. Tizzano

TW99. Quantitative analysis of SMN genes in Romanian spinal muscular atrophy (SMA) patients

M. Stavarachi, E. Zamba-Papanicolaou, A. Georghiou, P. Koutsou, C. Votsi,
P. Apostol, M. Toma, D. Cimponeriu, I. Radu, L. Gavrila

TW100. The zinc finger protein ZPR1 is a potential modifier of spinal muscular atrophy

L. Gangwan

TW101. Muscle hypertrophy in chronic spinal muscular atrophy: A case series

T. Mozaffar, K. Saremi, A. Wang

TW102. Role of proteasome in the formation of TDP-43 positive -
inclusions in spinal motor neurons in ALS

N. Kimura, T. Kumamoto, E. Ohama

S23

TW103. Orofacial characteristics of children with spinal muscular atrophy

D. Emmanouil

S29

TW104. Eosinophilia-Myalgia Syndrome following L-tryptophan ingestion

J. Allen, T. Al-Lahham, A. Peterson, R. Sufit, J. Varga

TW105. Autoimmune mechanism of sarcoid myopathy presenting as granuloma cells invasion into non-necrotic muscle fiber


TW106. Muscle pathology in diseases other than a true myopathies

J.M. Gray, A. Selva-O’Callaghan, S. Prieto-González, M. Butjosa, D. Martin,
M.A. Plasin, D. Cano, V. Lazo

TW107. Eosinophilic polymyositis and DRESS (drug rash with eosinophilia and systemic symptoms) syndrome associated with anti-tuberculosis drug

M.S. Park, J.Y. An, J.I. Seok, H. Noh

TW108. Molecular analysis of hibernation suggests novel treatment strategies to preserve muscle mass

E. Andres-Mateos, T.N. Burks, A. Soleimani, R. Marx, J.L. Simmers, B. Lin,

TW109. Immunohistochemical and electron microscopy study of myositis with rheumatoid arthritis

J. Shimizu, M. Hashimoto, M. Kadoya, Y. Koide, A. Kabota, S. Tsuji
TW110. Phosphorylation of ribosomal s6 kinases in atrophied and non-atrophied muscles of alcoholic patients is profoundly declined
B. Shenkman, E. Lysenko, O. Zinovyeva, G. Maslova, Y. Kazantseva, N. Yakhno

TW111. Historical perspective of Muscle Anatomy and Physiology through the eyes of Leonardo da Vinci
S.K. Mishra, O. Khayali, P. Singh

TW112. Interleukin-17 and Foxp3 expression in muscles from patients with Duchenne Muscular Dystrophy and Juvenile Dermatomyositis
A. D’Amico, L. De Pasquale, S. Petrini, F. De Benedetti, E. Bertini

TW113. In vitro studies of quiescent and reactivated human myoblast cultures
J. Sellathurai, S. Juhl Petersson, S. Cheedipudi, J. Dhawan, H. Daa Schroeder

TW114. The neurotrophin receptor p75NTR regulates human skeletal muscle cell differentiation and maturation
E. Colombo, S. Romaggi, E. Medico, M. Mora, C. Falcone, H. Lochmuller, P. Confalonieri, R. Mantegazza, L. Morandi, C. Farina

TW115. The neurotrophin receptor p75NTR is induced on mature myofibers in inflamed human skeletal muscle and promotes myotube survival to inflammatory stress
E. Colombo, S. Romaggi, F. Blasevich, M. Mora, C. Falcone, H. Lochmuller, P. Confalonieri, R. Mantegazza, L. Morandi, C. Farina

TW116. Prominent neuropathy in hereditary angioedema type III
M. Mahajan, M.D., Shalini; Engel, M.D., W. King

TW117. Integrating palliative care across the spectrum of life-threatening pediatric euromuscular conditions
C. Rushton, G. Geller, R. Cohn, L. Erby

TW118. Ethical challenges in the care of children and families affected by life-threatening neuromuscular disorders (LTNMDs)
G. Geller, C. Rushton

TW119. Overview of inflammatory myopathies in 2010: correlation between clinical, pathological and immunological data to an improved diagnosis
B. Fernandez, A. Maues De Paula, E. Salort-Campana, N. Schleinitz, A. Benyamine, P.J. Weiller, D. Figarella-Branger, F. Dignat George, J.F. Pellissier

Note: Posters numbered from TW01 to TW52 and from TW121 to TW158 will be discussed on Tuesday; Posters numbered from TW53 to TW119 and from TW159 to TW191 will be discussed on Wednesday.
h. 11.00 – 11.30  PL15.  The neuromuscular junction and congenital myasthenic syndromes  
*Hanns Lochmuller* (UK)

h. 11.30 – 12.00  PL16.  Muscle channelopathies – an altered electrical bistability of the fiber membrane  
*Frank Lehmann-Horn* (G)

h. 12.15 – 14.00  Lunch (self-served buffet)

h. 14.00 – 15.00  **MEET THE PROFESSOR -- 2 CONCURRENT SESSIONS**

**MTP7 (Red Hall) Motor Neuron Diseases**  
*L.P. Rowland* (Moderator) – *S. Appel – O. Hardiman – J. Melki – G. Tedeschi*

**MTP8 (Blue Hall) Limb-girdle Syndromes**  
*C. Angelini* (Moderator) – *K. Bushby – M. de Visser – V. Nigro – L. Politano- M. Zatz*

h. 15.00 – 17.00  **SIMULTANEOUS SYMPOSIA (S22-S28)**

**S22 (Hall E1) Myasthenic Syndromes**  
*Chairs: Angela Vincent and Henry Kaminski*

**Invited Speakers**

S22.L1. Neuromyotonia and muscle hyperexcitability: Clinical and electrophysiologic aspects  
*Kimiyoshi Arimura*

S22.L2. Current best therapies for Myasthenia Gravis  
*Amelia Evoli*

S22.L3. Clinical and epidemiological aspects of autoimmune myasthenic syndromes of autoimmune myasthenic syndromes  
*Jan Verschuuren*

**Platform Presentations**

S22.PP1.  *Congenital Myasthenic Syndrome (CMS) caused by mutations in Choline Acetyltransferase (ChAT). Clinical features, genetic analysis and expression studies in 11 patients*  

S22.PP2. *Late onset MG in the Northern Ireland population: an observed 10-fold increase in incidence from 1990-2008*  
*A. Carr, C. Cardwell, P. McCarron, D. O’Reilly, J. McConville*

S22.PP3.  *Psychosocial status of patients with myasthenia gravis and its impact on health-related quality of life*  
*I. Basta, S. Peric, V. Rakocvic-Stojanovic, Z. Stevic, I. Marjanovic, D. Lavrnic*

**S23 (Red Hall) Spinal Muscular Atrophy: Recent advances in clinical trials**  
*Chairs: Gideon Dreyfuss and Enrico Bertini*
Invited Speakers

S23.L1. Experience from clinical trials in SMA
Enrico Bertini

S23.L2. The use of SMA mice for development of therapies as well as understanding the basic mechanism of disease in spinal muscular atrophy (SMA)
Arthur Burghes

S23.L3. Biological outcome measure and validation in SMA
Francesco D. Tiziano

Platform Presentations

S23.PP1. Motor unit number estimation and compound motor action potential in spinal muscular atrophy patients: a prospective study

S23.PP2. Defining UBE-1 mutations and ubiquitination defects underlying X-linked lethal infantile SMA (XL-SMA)
L. Baumbach-Reardon

S24 (Blue Hall) Exercice therapy in neuromuscular diseases
Chairs: John Vissing and Ronald Haller

Invited Speakers

S24.L1. Exercise training in metabolic myopathies and mitochondrial diseases
Ronald G. Haller

S24.L2. Exercise therapy in CMT
Mary M. Reilly

S24.L3. Exercise therapy in muscular dystrophies
John Vissing

Platform Presentations

S24.PP1. Electrical stimulation and exercise therapy results in patients with limb girdle muscular dystrophy: A controlled clinical trial
M. Kilinc, S. Aksu Yildiryim, S. Atay Yilmaz, E. Tan

S24.PP2. Neuromuscular electrical stimulation training: a safe and effective treatment for facioscapulohumeral muscular dystrophy patients
S. Sacconi, S. Colson, V. Tanant, M. Benchortane, C. Benaim, M. Fournier-Mehouas, C. Desnuelle

S24.PP3. Effect of aerobic training in patients with spinal muscular atrophy type III
R.S. Hansen, K.L. Madsen, F. Thoegersen, N. Preisler, M. Berthelsen, J. Vissing
S25 (Hall E2)  
Friedreich’s Ataxia  
Chair: Feza Deymeer and Alessandro Filla

Invited Speakers

S25.L1. The dominant ataxias  
Alessandro Filla

S25.L2. Differential diagnosis of the newly discovered recessive ataxias  
Michel Koenig

S25.L3. Hereditary spastic paraplegias  
Goekhan Uyanik

Platform Presentations

S25.PP1. Investigation of a Cypriot autosomal recessive cerebellar ataxia family linked to 9p: evidence for a new locus?  
C. Votsi, E. Zamba-Papanicolaou, M. Pantzaris, K. Christodoulou

S25.PP2. Effects of erythropoietin on neovascularisation and metabolism of skeletal muscle in Friedreich Ataxia  
W. Nachbauer

S26 (Hall F1)  
Animal models for neuromuscular diseases  
Chair: Vincenzo Nigro and Diane Shelton

Invited Speakers

Jeffrey Chamberlain

S26.L2. Investigating valproate as a treatment for McArdle’s disease  
John Mc Howell

S26.L3. The BIO14.6 hamster as a model for long life gene therapy of cardiomyopathy and muscular dystrophy  
Vincenzo Nigro

S26.L4. Mice models deficient for two muscle proteins helping to understand protein complexes organization and function  
Mariz Vainzov

Platform Presentations

S26.PP1. Mono and di-allelic titinopathy in a mouse model carrying the c-terminal FINmaj titin mutation  
S26.PP2. Etanercept suppressed the necrosis and inflammatory change in muscle of SJL/J mice by blocking TNF α function  

S27 (Hall F2)  
Outcome measures in neuromuscular diseases  
Chairs: Marianne de Visser and Eugenio Mercuri

Invited Speakers

S27.L1. Outcome measures in paediatric patients  
Eugenio Mercuri

S27.L2. Outcome measures in peripheral neuropathies  
Davide Pareyson

S27.L3. Outcome measures in adults with muscle diseases  
Michael R. Rose

Platform Presentations

E. Henricson, C. McDonald, R.T. Abresch, J. Han, R. Leshner, A. Cnaan, F. Hu, A. Zimmerman, T. Duong, A. Arrieta, The CINRG Investigators

S27.PP2. The Motor Function Measure (MFM) in the assessment of Pompe disease  
D. de Castro, C. Payan, K. Laloui, A. Canal, P. Laforêt, C. Bérard, The French Pompe Register, Study Group

S27.PP3. MRI in LGMD2I; a qualitative and quantitative analysis using the 3 point Dixon technique  
T. Willis, K. Hollingsworth, A. Mayhew, M. Eagle, K. Bushby, H. Lochmuller, V. Straub

S28 (Hall F3)  
New therapies in muscle channelopathies  
Chairs: Reinhardt Rüdel and Georges Serratrice

Invited Speakers

S28.L1. Therapy of episodic weakness or neuropathic pain  
Diana Conte Camerino

S28.L2. Novel aspects of diagnosis and therapy in the chloride channel myotonias  
Frank Lehmann-Horn

Michael G. Hanna
Platform Presentations

S28.PP1. Newly synthesized pyrroline derivatives of mexiletine as a potentially dually acting sodium channel blockers and anti-oxidant compounds with wider therapeutic application in neuromuscular disorders
M. De Bellis, V. Sblandorio, A. Carocci, M.M. Cavalluzzi, G. Lentini, C. Franchini, A. De Luca, D. Conte Camerino

S28.PP2. Successful flecainide therapy in Paramyotonia congenita
O. Farina, D. Di Diodato, G. Ciccone, MS Mayer, S. Sampaolo, G. Di Iorio

h. 17.00 – 18.30 POSTER PARALLEL SESSIONS (14 – 17)

POSTER SESSION 14:
GENE THERAPY IN MUSCULAR DYSTROPHIES & OUTCOME MEASURES IN MUSCULAR DISEASES
Chairpersons: S. Braun, E. Mercuri

TW121. Heparin-binding influences tissue tropism of AAV1 and AAV6 and increases efficiency of striated muscle transduction in wt mice
A. Arnett, L. Beutler, A. Quintana, J. Allen, E. Finn, R. Palmier, J. Chamberlain

TW122. Codon-optimized AAV vector for gene therapy in BIO14.6 hamsters
L. Rotundo, A. Lancioni, G. Nigro, D. Di Napoli, S. Castaldo, A. Auricchio, G. Piluso, V. Nigro

TW123. Systemic transplantation of Human Adipose-Derived Stem cells into the Golden Retriever Dystrophic dog

TW124. Lesson from the patients skipped by nature: characterization of Becker Muscular Dystrophy (BMD) with Exon 51 as one endpoint
S. Cirak, V. Arechavala-Gomeza, L. Feng, M. Guglieri, S. Torelli, M. Kinali, J.E. Morgan, C. Sewry, K. Bushby, A. Ferlini, F. Muntoni

TW125. Antisense RNA/ethylene-bridged nucleic acid chimera induces exon 45 skipping and restores dystrophin expression in DMD muscle cells
M. Yagi, M. Ota, H. Awano, Y. Takeshima, M. Matsuo

TW126. Long-term administration of antisense oligonucleotide against dystrophin exon 19 for the treatment of Duchenne muscular dystrophy with exon 20 deletion
M. Matsuo, Y. Takeshima, M. Yagi, H. Awano

TW127. Potential role of PAX7 transfected mesenchymal stem cells in the treatment of Duchenne Muscular Dystrophy


TW129. Restoring cell-basal lamina interaction to rescue tissue degeneration in congenital muscular dystrophy

TW130. The bone marrow mesenchymal stem cells can rescue damaged skeletal muscle
TW131. Outcome measures validation study for mesoangioblasts transplantation in children affected by Duchenne Muscular Dystrophy

TW132. The OBFR score complements the MG composite and the MG-QOL15
M.E. Farrugia, H. Harle, T. Burns

TW133. A quantitative measure of hand grip myotonia in non-dystrophic myotonias
J. Statland, Y. Wang, B. Bundy, J. Trivedi, L. Herbelin, S. Pandya, W. Martens, R. Barohn, CINCH Consortium

TW134. Interactive voice response diary: A patient-reported outcome measure in nondystrophic myotonias

TW135. Twelve-month interim results of an open label extensions study (DELPHI-E) with idebenone (Catena®) in Duchenne Muscular Dystrophy (DMD)
G.M. Buyse, N. Goemans, M. Van den Hauwe, D. Thijs, L. Mertens, T. Meier

TW136. Assessment of tongue pressure during swallowing in patients with muscular dystrophies
T. Matsumura, S. Hamanaka, J. Kondo, K. Hori, T. Ono, H. Fujimura, S. Shinno

TW137. Baseline and one-year pulmonary function data of boys with severe dystrophinopathies
R.T. Abresch, C. McDonald, E. Henricson, J. Han, R. Leshner, A. Cnaan, A. Zimmerman, CINRG Investigators

TW138. Parent proxy-reported health-related quality of life in an observational study of boys with confirmed Duchenne muscular dystrophy using the PedsQL generic core scales
E. Henricson, C. McDonald, R.T. Abresch, J. Han, R. Leshner, A. Cnaan, A. Zimmerman, A. Adrienne, CINRG Investigators

TW139. Analysis of different types of mononeuropathies with regard to it’s causes and the treatment outcomes
S. Misevic, B. Kovac, D. Vukasinovic-Soljacic, L. Knezevic-Poljak, S. Tomic, S. Butkovic-Soldo, B. Kovac

TW140. Bilateral phrenic nerve injury following endoscopic ablation for atrial fibrillation
L. Boissé, K. Kimpinski, P. Bourque, C. Bolton

POSTER SESSION 15:
OUTCOME MEASURES IN MUSCULAR DISEASES
Chairpersons: M. Rose, D. Pareyson

TW141. Causes and outcome of acute neuromuscular respiratory failure
M. Cabrera Serrano, A. Rabinstein

TW142. Amyotrophic Lateral Sclerosis (ALS) and Cervical Spondylosis/Cervical Spondylotic Myelopathy (CS/CSM): experience in a Referral Centre
G. Gargiulo Monachelli, G. Rodriguez, M. Bettini, S. Rey, R. Sica

TW143. Epidemiological, demographic and occupational characteristics of Amyotrophic Lateral Sclerosis patients in a referral centre
M. Bettini, G. Gargiulo Monachelli, G. Rodriguez, R. Sica, R. Rey

TW144. Back to the basis; a forgotten reflex in a motor neuron disease
H.D. Gonorazky, S. Rodriguez, M. Bettini, D. Consalvo, G. Rodriguez
TW145. Registries and their developing role in outcomes research for rare childhood diseases: A review of current literature
S. Prasad, E. James, S. Jones

TW146. Factors associated with taste abnormalities in patients with subacute myelo-optico-neuropathy (SMON)

TW147. A reduced version of the Motor Function Measure scale (MFM) for young children with Neuromuscular Disorders: Reliability and construct validity
C.A.M. Payan, C. de Lattre, C. Berard, MFM-20 Study Group

TW148. TREAT-NMD Registry of Outcome Measures (ROM); finding what you need fast
Auld, Jo; Rose, Michael; Seyedsadjadi, Reza

TW149. Validation of Serbian version of the Individualized Neuromuscular Quality of Life questionnaire (INQoL) in patients with myotonic dystrophy type 1
S. Peric, V. Rakovec-Stojanovic, I. Basta, M. Miljkovic, I. Marjanovic, D. Lavrnic

TW150. Quantitative muscle ultrasonography as a follow-up tool in Duchenne Muscular Dystrophy: validation with clinical measurements
M. Jansen, N. van Alfen, M.W.G. Nijhuis- van der Sanden, S. Pillen, I.J.M. de Groot

TW151. Manual muscle test of the upper limbs. A way of measuring muscle function in Spinal muscular atrophy II?
U. Werlauff, S. Berthelsen, I. Fløytrup, B. Kristensen, B. Steffensen, B. Werge

TW152. Psychometric evaluation of the Health Assessment Questionnaire (HAQ) and its domains in patients with muscle disease in US and UK populations
M. Rose, R. Sadjadi, H. Al-Sarraj, Muscle Study Group, MSG

TW153. Receiver and transmit non-uniformity correction: a mandatory step towards muscle quantitative NMR imaging
N. Azzabou, P. Loureiro da Sousa, P. G. Carlier

TW154. The Scottish muscle managed clinical network: A model for equitable service provision

TW155. New challenges in the management of neuromuscular disorders: the need for a “medicine” of neuromuscular disorders

TW156. Reproducibility and reliability of quantitative MRI measures for neuromuscular disease

TW157. Quantification of the upper-limb functionality during daily activity in patients with neuromuscular diseases: A pilot study
R. Ganea, K. Aminian, A. Paraschiv-Ionescu, I. Virnot, P.Y. Jeannet

TW158. Quantitative magnetization transfer MRI: A potential new source of biomarkers in skeletal muscle?

POSTER SESSION 16:
CONGENITAL MYOPATHIES
Chairpersons: C. Wallgren-Pettersson, N. Romero

TW159. Atypical or unfrequent muscular disorders in neuropediatric syndromes
G.T. Serratrice, B. Chabrol
TW160. Analysis of the nebulin gene by multiplex-liquidation dependent probe amplification (MLPA) and methylation-specific MLPA – an update (withdrawn)
M. Lunkka-Hytonen, V.L. Lehtokari, C. Wallgren-Pettersson, K. Pelin

TW161. Congenital myopathy with type 1 fiber uniformity associated with a mutation in the beta tropomyosin gene, TPM2
H. Taishzarghi, L. Palm, A. Oldfors

TW162. Respiratory impairment, deformity of the spine and low body mass index are major features in juvenile Selenoprotein N1 – related myopathy (SEPN1-RM)

TW163. New data on a case of congenital fibre type disproportion and non compaction cardiomypathy associated with insulin resistance
D. Diodato, S. Sampaolo, A. Varone, G. Limongelli, M. Simonetti, P. Calabrò, R. Calabrò, G. Di Iorio

TW164. Congenital myopathy with moyamoya syndrome and schwannoma
T. Fujii, T. Miyajima, T. Kamada, N. Kimura, H. Shimomura, K. Saito

TW165. Selenoprotein-N (SEPN1) congenital muscular dystrophy with total COX depletion and multi-mini cores – expanding the morphologic spectrum
C. D’Arcy, M. Ryan, E. Yiu, L. Shield, C. McLean

TW166. Clinical and genetic characterization of patients with merosin-deficient congenital muscular dystrophy
Y.E. Park, N.Y. Jung, H.S. Kim, Y.B. Shin, D.S. Kim

TW167. Clinical and pathological variability of congenital myopathies associated with recessive mutations in the RYR1 gene

TW168. Epidermolysis bullosa with late-onset muscular dystrophy and plectin deficiency

TW169. Congenital muscular dystrophies: a clinical and histopathological study
M. Nagappa, N. Atchayaram, G. Narayanappa

TW170. Nemaline myopathy type 6: clinical and myopathological features
M. Olive, L. Goldfarb, H.S. Lee, O. Zagaa, A. Blokhin, L. Gonzalez-Mera, D. Moreno, N. Laing, S. Nyamkhishig

TW171. Over expression of synemin improves the desmin network in desminopathy
O. Chourbagi, F. Bruston, Z. Xue, Z. Li, P. Vicart, O. Agbulut

TW172. Mutational analysis of CLCN1 and SCN4A genes in Czech patients with non-dystrophic myotonia
J. Sedlackova, D. Paclova, S. Vohanka, L. Fajkusova

TW173. Scoliosis Surgery (ScSu) in respiratorily-compromised congenital neuromuscular patients can fatally worsen ventilatory function or engender nocturnal BiPAP dependency, as evidenced in four children with Focal-Loss-of-Cross-Striations (FLCS)
S. Mahajan, W.K. Engel

TW174. Scoliosis surgery in a patient with hyaline body myopathy

TW175. Myopathy with tubular aggregates related to malignant hyperthermia: A new clinical group?
A. Maues De Paula, C. Fernandez, S. Attarian, JF Pellissier, J. Pouget, D. Figarella-Branger
POSTER SESSION 17:  
MISCELLANEOUS  
Chairpersons: M. Moggio, G. Siciliano

S29  
TW176. Non-muscle myosin II-C is abundantly expressed in skeletal muscle and associated with Z-lines  
C. Terracciano, E. Lena, A. Botta, G. Bernardi, R. MASSA

TW177. Acquired myopathy induced by statin and fibrate in skeletal muscle: a proteomic and genomic approach  
G. Camerino, S. Pierro, C. Digenarro, M.A. Pellegrino, R. Bottinelli, A.L. George, D. Conte Camerino

TW178. Role of thrombospondin-1 in macrophage inflammation in dysferlin myopathy  
N. De Luna, E. Gallardo, C. Sonnet, B. Chazaud, X. Suarez-Calvet, R. Gherardi, I. Illa

TW179. Macrophagic myofasciitis-associated chronic dysfunction  

TW180. MuRF-1 gene deletion prevents atrophy of disused muscle in the hindlimb-unloaded mice  
J.F. Desaphy, S. Pierro, C. Digenarro, G.M. Camerino, M. Sandri, S. Schiaffino, D. Conte Camerino

TW181. Wound botulism in drug users: a still underestimated diagnosis  
E. Barca, P. Girlanda, L. Fencia, F. Anniballi, U. Sinardi, C. Rodolico, A. Toscano

TW182. Acquired multifocal myokymia-myoclonus – Case histories, light – and electron microscopy of muscle biopsies  

TW183. A hug for all children: A public awareness program changing knowledge and attitudes of elementary school children towards children with neuromuscular disorders  
D. Emmanouil, A. Smerou, A. Terzidis

TW184. The incidence of adverse reactions in patients with neurological disorders receiving plasmapheresis treatment  
B. Givenc, F. Koc, F. Tekinturhan, D. Turgay, S. Turgut, M. Karadamar, I. Unal

TW185. Type I interferons and Toll-like receptors: a new insight into the etiology of idiopathic inflammatory myopathies  

TW186. CRYAB mutation causing multisystemic disease affect residues that interact during (alpha)B-crystallin dimerization  

C. McDermott, G. Chavada, A. Al-Nayal, F. Lee, S. Webber, M. McAlindon, T. Walsh, H. Hollinger, P. Shaw

TW188. Distribution of charcot-marie-tooth disease and frequency of positive genetic testing results  

TW189. Serological inflammatory profile in LGMD2B: good or bad macrophage-muscle fiber interaction?  
C. Borsato, S. Cagnin, L. Bortolussi, M. Fanin, C. Angelini
TW190. Characterization of altered processes and protein modifications in patients affected by inflammatory myopathies

TW191. Hippocampal atrophy related to memory performance in non-demented ALS patients: a voxel based morphometry and neuropsychological study
J. Raaphorst, M.J. van Tol, M. de Visser, W. Linssen, L. van den Berg, B. Schmand, D. Veltman

Note: Posters numbered from TW01 to TW52 and from TW121 to TW158 will be discussed on Tuesday; Posters numbered from TW53 to TW119 and from TW159 to TW191 will be discussed on Wednesday.

h. 18.30 – 19.00 Poster Removal
h. 20.30 Gala Dinner – Naples Archaeological National Museum

Wednesday – July 21st, 2010

h. 9.00 – 17.00 WANDA Plenary Session (Hall F5)
Chairs: Anna Ambrosini (Italy), Allan Bretag (Australia), Ysbrand Poortman (Netherlands)

h. 9.00 – 10.30 Worldwide support for NMD Treatment/Prevention
Reports from WANDA regional representatives

h. 10.30 – 10.50 Coffee break

h. 10.50 – 13.00 The Mediterranean Situation: Opportunities, Barriers
Invited presentations
Forum: The needs in the Mediterranean area

h. 14.00 – 15.00 From National Activities to Global Joint Ventures
Tools

h. 15.00 – 17.00 WANDA/ICNMD joint session: PARTNERING RESEARCH AND SOCIETY
h. 15.00 – 15.30 WANDA – The Road Map to Treatment in NMD
h. 15.30 – 16.00 TREAT-NMD – Achievements and Future Perspectives
h. 16.00 – 16.30 ENMC – A platform of Empowered Patient Organisation
h. 16.30 – 17.00 AOMC – Education and Training

h. 17.00 – 17.15 Coffee break

h. 17.15 – 19.00 WANDA Assembly (Red Hall)
Summary of ICNMD highlights
Fundraising by Telethon
Reports from National NMDAs
Thursday – July 22nd, 2010

h. 8.00 – 10.00  Poster removal, coffee and rolls

h. 8.00 – 10.00  WANDA Assembly continued (Blue Hall)
Business meeting
Reports from National NMDAs continued (if necessary)

h. 10.00 – 12.00  PLENARY LECTURES (Red and Blue Halls)
Chairs: Hans Goebel (Germany) and Louis Kunkel (USA)

h. 10.00 – 10.30  PL17. Mechanistic and molecular insights into the pathogenesis of muscular dystrophy
Kevin Campbell (USA)

h. 10.30 – 11.00  PL18. State of the art in laminopathies
Giuseppe Novelli (I)

h. 11.00 – 11.30  PL19. Autoimmune inflammatory myopathies
Marinos Dalakas (UK)

h. 11.30 – 12.00  PL20. State of the art in Friedreich’s ataxia
Massimo Pandolfo (B)

h. 12.15 – 14.00  Lunch (self-served buffet)

h. 14.00 – 15.00  MEET THE PROFESSOR -- 2 CONCURRENT SESSIONS

MTP9 (Red Hall) Metabolic Myopathies
S. Di Mauro (Moderator) – Z. Argov – C. Desnuelle – I Nishino – M. Zeviani

MTP10 (Blue Hall) Inherited Neuropathies
I. Hausmanowa-Petrusewitz (Moderator) – G. Di Iorio – M. Reilly – M. Shy – G. Vita

h. 15.00 – 16.00  LATE BREAKING NEWS SESSION (Red and Blue Halls)
Chairs: Valerie Askanas (USA) and Luisa Politano (Italy)

Oral Communications from Selected Late Breaking News Abstracts

h. 16.00 – 17.00  Summary of Scientific Highlights

Presenters:
Stanley Appel: Motor Neuron Diseases
Michael Shy: Peripheral Nerve Diseases
Angela Vincent: Neuromuscular Junction Diseases
Francesco Muntoni: Muscle Diseases

h. 17.00 – 17.30  Handover of the ICNMD to the next host and closing ceremony

h. 20.00  “Arrivederci” Dinner - Donn’Anna Palace, Posillipo
General Information

Language
Official language will be English.

Congress Venue: The Congress will take place in the Monte S. Angelo University Campus – Fuorigrotta, a large and comfortable Congress Centre, not far from the city centre. Opening Ceremony and Welcome Dinner will take place in the garden of the Royal Palace Theatre. Gala Dinner will take place in the National Archaeological Museum.

Shuttle Service
A shuttle service will be available from the hotels to the Congress venue and viceversa, during the entire period of the Congress.

Registration Info
Online registration is available from September 15, 2009

Registration fees

Active participants:
Payment received by March 31\textsuperscript{st}, 2010: € 600
Payment received after March 31\textsuperscript{st}, 2010: € 750

Persons under 35 years of age:
Payment received by March 31\textsuperscript{st}, 2010: € 320
Payment received after March 31\textsuperscript{st}, 2010: € 700

Accompanying persons:
Payment received by March 31\textsuperscript{st}, 2010: € 220
Payment received after March 31\textsuperscript{st}, 2010: € 220

WANDA delegates:
Payment received by March 31\textsuperscript{st}, 2010: € 320
Payment received after March 31\textsuperscript{st}, 2010: € 700
One-day registration (Wednesday 21\textsuperscript{st} July): € 100

Teaching courses participants:
Payment received by March 31\textsuperscript{st}, 2010: € 100
Payment received after March 31\textsuperscript{st}, 2010: € 200

The Congress fee includes: bus transportation, welcome reception, coffee breaks, lunches, Gala Dinner*, Congress book and program, Certificate of attendance.

*The participation to the Gala Dinner requires an additional fee of € 50,00 and it is limited to 500 persons.

Deadlines
March 1\textsuperscript{st}, 2010: Submission of Abstracts
May 31\textsuperscript{st}, 2010: Submission of Late Breaking News Abstracts

Prizes
The best platform or poster presentations by a young researcher – still in training – will awarded by prizes of 500 Euros each, according to the decision of an international multidisciplinary panel of judges. It is at discretion of the committee to select either poster or oral presentation.
CME Credit Program

E.C.M.
The Italian National Commission for continues Training
With reference to the request for accreditation No event 1344-10020034 entitled “12TH INTERNATIONAL CONGRESS ON NEUROMUSCULAR DISEASES” has given 15 (fifteen) credits.

La Commissione nazionale per la formazione continua (E.C.M.)
In riferimento alla richiesta di accreditamento dell’evento n. 1344-10020034 dal titolo “12TH INTERNATIONAL CONGRESS ON NEUROMUSCULAR DISEASES” i crediti formativi proposti dalla Commissione nazionale per la formazione continua sono 15 (quindici).

Climate
The climate in Naples in July is usually sunny, with average temperatures varying between 25 and 30°C.

Naples Environ
Capri, Ischia, Sorrento, Amalfi, Pompei, Ercolano, Vesuvius, Caserta, Positano, Campi Flegrei

Information
Congrex – a leading international management company, offering comprehensive services for meetings, events, conferences, association management, travel and accomodation – is the official organizer of the ICNMD 2010.

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website: www.icnmd2010naples.org
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Have you ruled out Pompe Disease?
The management of Pompe disease differs from other neuromuscular diseases in that specific treatment is now available, making early recognition of the disease a priority. 1

Confirmatory Diagnosis
Can be made quickly and accurately in blood (dried blood spots, lymphocytes or leukocytes). 2, 3

For more information on the use of Dried Blood Spot Samples to diagnose Pompe Disease, please come and see us at stand nr. 1 & 2.
Have you ruled out Pompe Disease?

Confirmatory diagnosis can be made quickly and accurately in blood (dried blood spots, lymphocytes or leukocytes) 1, 2, 3, 4

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