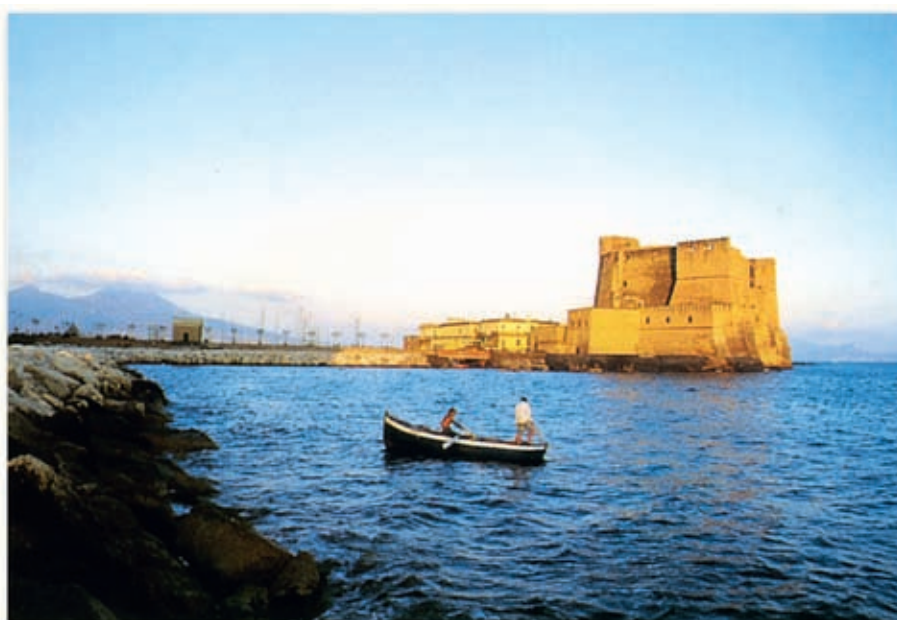


World Federation of Neurology

XII International Congress

on Neuromuscular Diseases

in Memory of Lucia Ines Comi



Et in Arcadia ego... Auch in Arcadia...
WOLFGANG GOETHE (Letters from Naples)

Naples, Italy

July 17th-22nd, 2010

PROGRAM

President of the Congress

Giovanni Nigro

Honorary Presidents

Salvatore Di Mauro, W. King Engel

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Raffaele Calabrò, Roberto Cotrufo

Honorees List

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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, pathogenesis 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

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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
Unione Italiana Lotta alla Distrofia Muscolare

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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 **Rbdomyolysis: 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.L1. Mitochondrial Dysfunction of Aging Skeletal Muscle: A potential therapeutic Focus**
David A. Hood
- S1.L2. 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

A. Kochoński, D. Kabzińska, H. Strugalska-Cynowska, A. Kostera-Pruszczyk, B. Ryniewicz, R. Posmyk, A. Midro, P. Seeman, L. Barankova, M. Zimoń, J. Baets, A. Jordanova, V. Guergueltcheva, I. Tournev, S. Sarafov, P. De Jonghe, V. Timmerman, I. Hausmanowa-Petrusewicz

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

P.Laforêt, C. Acquaviva-Bourdain, N. Streichenberger, S. Attarian, A. Labarre-Vila, C. Vanhulle, A. Laquerrière, G. Bassez, L. Pérard, P. Kaminsky, O. Rigal, M. Brivet, J. Pouget, B. Eymard, C. Vianey-Saban

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-Pruszyk, 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 Petrusiewicz

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

J. Gurgel-Giannetti, M. Silva, Z. Meira, R.A. Ferreira, L.U. Yamamoto, M. Vainzof, M. Zatz, C.E. Rochite

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

h. 18.00 – 19.30 **POSTER PARALLEL SESSIONS (1 – 5)**

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. Flaibani, 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. Jønck

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. Kilinç, S. Aksu Yıldırym, S. Atay Yılmaz, E. Tan

SM15. The effect of body composition on motor performance in adult patients with neuromuscular diseases

S. Aksu Yıldırym, M.E.Yapa, E. Tan

POSTER SESSION 2:

CARDIOMYOPATHIES & NUCLEAR ENVELOPE DISEASES

Chairpersons: G. Bonne, G. Nigro

S6

SM16. Dilated cardiomyopathy associated with mutations in delta-sarcoglycan gene. Report of two familial cases

L. Passamano, A. Palladino, E. Viggiano, P. Ambrosio, G. Piluso, V. Nigro, L. Politano

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

L. Bello, R. Pezzani, J. Smaniotta, A. D'Amico, E. Bertini, E. Leonardi, S. Tosatto, C. Angelini, E. Pegoraro

SM19. Left ventricular diastolic function in Myotonic Dystrophy

A. Palladino, Ge. 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

M.Orlandi, C. Giordano, M.G. Bates, G. Palano, E. Perli, C. Nediani, D. Turnbull, R.W. Taylor, G. d'Amati

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

S14

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

SM26. Novel start codon mutation in the lamin A/C gene: clinical and pathological findings

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

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

C. Magagnotti, I. Fermo, S. Previtali, G. Zerbini, D. Gabellini, E. Fattore, M. Ferrari, A. Bachi, A. Andolfo, S. Benedetti

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. Companys, 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 Blecker, 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. Tsujii***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)***Y. Inamori, I. Higuchi, R. Ohkubo, N. Hirata, A. Hashiguchi, K. Higashi, T. Shiraishi, H. Takashima***SM36. Expression of VCP in muscle fibers of experimental chloroquine-induced myopathy***Y. Aso, K. Sawada, T. Masuda, Y. Hazama, K. Nakamura, N. Kimura, T. Kumamoto***SM37. Dysregulation of myogenic regulatory factors and myo-endothelial remodelling in sporadic inclusion body myositis***J. Wanschitz, O. Dubourg, E. Lacene, M. Fischer, R. Hoeflberger, H. Budka, N.B. Romero, B. Eymard, S. Herson, G. Butler-Brown, T. Voit, O. Benveniste***SM38. Prevalence of inclusion body myositis (IBM) in Japanese population***N. Suzuki, M. Aoki, M. Tateyama, R. Izumi, T. Akiyama, H. Warita, Y. Itoyama, M. Mori, H. Kusaka, I. Higuchi, T. Kondo, M. Uchino, R. Kaji, I. Nishino***SM39. Magnetic Resonance Imaging of skeletal muscles in sporadic inclusion body myositis***F. Cox, M. Reijniersie, 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. Rosenzweig, S. Herson, 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. Ohara, 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. Wanschitz, M. Rosenzwajg, 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. Kirschner

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. Camaño, 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. Patrino, 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

Ge.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. Calabrò, L. Politano

SM52. Relationships among organ involvements in myotonic dystrophy

P. Kaminsky, M. Poussel, L. Pruna, B. Chenuel, 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. Perdoni, 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 I

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. Siritto, 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***Ge.Nigro, V. Russo, L. Delli Paoli, A.A. Papa, A. Merola, F. Di Meo, A. Palladino, M.G. Di Gregorio, M.G. Russo, P. Golino, R. Calabrò, L. Politano***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. Zaoralikova, 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 Vazquez, R.E. Escobar***SM73. CARE-NMD: Dissemination and implementation of standards of care for Duchenne Muscular Dystrophy in Europe (including Eastern Europe)***K. Gramsch, A. Tassoni, S. Geismann, J. Vry, R. Korinthenberg, J. Kirschner, H. Lochmüller, J. Rahbek, O. Gredal, K. Bushby, V. Straub, R. Thomson, I. Tournev, V. Guergueltcheva,*

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. Sorarù, 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. Gazzero, 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. Tasca, M. Pescatori, M. Mirabella, M. Monforte, T. Cubeddu, E. Iannaccone, R. Frusciantè, 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?

F. Greco, I. Frambolli, C.Borsato, M. Cao, L. Vercelli, M. Servida, L. Peverelli, L. Colantoni, G. Ricci, L. Volpi, R. Frusciantè, I. Scionti, M. Govi, E. Bonifazi, C. Forghieri, R. Di Leo, C. Manzoli, P. Cudia, E. Pastorello, L. Ricciardi, L. Morandi, C.P. Trevisan, G. Siciliano, G. Galluzzi, E.Ricci, A. Di Muzio, C. Rodolico, G. Tomelleri, M. Moggio, L. Palmucci, C. Angelini, L. Santoro, R. Tupler

SM84. About the facioscapulohumeral 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

L. Papić, D. Fischer, S. Trajanoski, R. Höftberger, C. Fischer, T. Ströbel, R. Bittner, M. Schabhüttl, K. Gruber, T. Pieber, A. Janecke, M. Auer-Grumbach

SM88. Mutation analysis of Limb girdle muscular dystrophies in the Czech Republic

Fajkusova, Lenka; Stehlikova, Kristyna; Vondracek, Petr; Hermanova, Marketa

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. Sundaram²; S.M. Uppin, J. Afshan, R. Mridula, LSR Krishna, R. Borgohain

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

C. Paradas, J. Llauger, J.Díaz-Manera, R. Rojas-García, N. De Luna, C. Iturriaga, C. Márquez, M. Usón, K. Hankiewicz, E. Gallardo, I. Illa

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 ANOS1/TMEM16E

S. Penttilä, O. Raheem, T. Suominen, M. Rötttä, P. Hackman, B. Udd

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

L. Volpi, G. Ali, S. Linsalata, M.Tosetti; R. Battini, G. Astrea, G. Ricci, V. Calsolaro, E. Caldarazzo Ienco, C.Nesti, L. Petrozzi, M. Mancuso, L. Pasquali, M. Fanin, C.Angelini, A. Pini, G. Fontanini, G. Cioni, G. Siciliano

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

J. Sarparanta, G. Blandin, K. Charton, A. Vihola, S. Marchand, A. Milic, P. Hackman, E. Ehler, I. Richard, B. Udd

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

P.C.Martins-Machado, D. Ayub-Guerrieri, P. Onofre-Oliveira, G. Monteiro, A.F. Santos, D. Zilbersztajn-Gotlieb, L.E.S. Netto, M. Vainzof

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. Vorgerd

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-oculopharyngeal 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 (HMERF) 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. Proteomic 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

J.C. Van den Bergen, A.J. Van der Kooi, E.H. Niks, C.S.M. Straathof, A.M. Aartsma-Rus, G.J. Van Ommen, J.J.G.M. Verschuuren, H.B. Ginjaar

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

F. Pinto-Mariz, L. Carvalho, W. de Mello, A. Araújo, M. Ribeiro, M. Cunha, I. Riederer, E. Negroni, T. Voit, I. Desguerre, G. Butler-Browne, W. Savino, S. Silva-Barbosa

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

M. Fardeau (Moderator) – V. Askanas – W.K. Engel – A. Oldfors – C. Sewry

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

O. Benveniste, M. Guiguet, J. Freebody, S. Herson, O. Dubourg, W. Squier, I. Leite, B. Eymard, D. Hilton-Jones

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 (DMI)
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 OPA1 gene
M.L. Valentino, L. Caporali, R. Liguori, P. Barboni, M. Carbonelli, R. Carroccia, L. Iommarini, A. Maresca, C. La Morgia, P. Avoni, P. Martinelli, A. Baruzzi, C. Lamperti, E. Lamantea, M. Zeviani, P. Montagna, V. Carelli

S10.PP2. MELAS and L-arginine therapy. Five years outcome on 8 MELAS patients
Y. Koga, N. Povalko, K. Katayama, N. Kakimoto, J. Nishioka, T. Matsuishi

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

S11.L3. Morphological aspects in extracellular matrix myopathies

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

S12.PP2. Motor chip: a CGH microarray for neuromuscular disorders

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

S13.L3. Modifiers of motor neuron degeneration

Wim Robberecht

Platform Presentations

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

S. Corti, M. Nizzardo, M. Nardini, C. Donadoni, S. Salani, C. Simone, M. Falcone, G. Riboldi, N. Bresolin, G. Comi

S13.PP2. Corpus callosum DTI as a biomarker for amyotrophic lateral sclerosis

N. Filippini, C. Mackay, G. Douaud, S. Knight, K. Talbot, M. Turner

S13.PP3. Adult neural stem cells from spinal cord of G93A: a new in vitro model of Amyotrophic Lateral Sclerosis

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 alpha 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^{-/-}/Lmna^{H222P/H222P} mice: focus on the heart

W. Liang, Y.K. Hayashi, Y. Park, H. Mitsuhashi, T. Arimura, G. Bonne, S. Noguchi, I. Nishino Ichizo

S14.PP3. Mutation of SYNE-1, encoding an essential component of the nuclear lamina, is responsible for autosomal recessive arthrogyrosis

R. Attali, N. Warwar, A. Israel, I. Gurt, E. McNally, M. Puckelwartz, B. Glick, Y. Nevo, Z. Ben-Neriah, J. Melki

h. 17.00 – 18.30 **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

E. Yiu, N. Geevasinga, G. Nicholson, E. Fagan, M. Ryan, R. Ouvrier

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. Altin, G. Vyshka, J. Kruja

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

A. Vital, Ph. Latour, G. Sole, C. Goizet, X. Ferrer, C. Vital, F. Tison

SM129. The phenotype of the Gly94fsX222 PMP22 insertion

S. De Vries, F. Van Ruissen, C. Verhamme, W.F. Arts, H. Mauser, H. Kerkhoff, B. Van Engelen, M. Lammens, M. De Visser, F. Baas, A. Van der Kooij

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 desoxysphingoid bases and disease severity in HSN1

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. Jyothisna, S. Aparna, R. Renuka, M.S. Lavanya, M.D. Sadhanani

SM140. Mosaicism for GJB1 (Cx32) mutation causes milder Charcot-Marie-Tooth typ X1 (CMTX1) phenotype in heterozygous man than in full carrier woman

P. Seeman, R. Mazanec, I. Sakmaryová, E. Kožárek, L. Baránková, M. Havlova

SM141. Charcot Marie Tooth Disease and Neuromyotonia

Y. Parman, E. Shugaiv, Z. Matur, M. Poyraz, P. Serdaroglu-Oflazer, 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

S2

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 neuro-lalic amyotrophy- expansion of the phenotype*D. Lev, T. Sagie, M. Sadeh, E. Lyshinsky-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. Kuruvilla, V. Bril***SM155. Acute tetraparesis in a pregnant woman as onset of an acute intermitent porphyria***M.A. Rubio Perez, J. Pascual Calvet, M. Sepulveda Gazquez, E. Giralt Steinhauer, S. Jimena Garcia, L. Planellas Gine, E. Munteis Oliva, J. Roquer Gonzalez***SM156. Prevalence of tarsal tunnel syndrome in patients with fibromyalgia: comparison with normal controls***S.J. Na, S.C. Lee, C.I. Joung, K.O. Lee***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. Khorvash***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. Vlckova-Moravcova, M. Nemecek, 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. Haberlová, R. Mazanec, Z. Rychlý, J. Sabová, J. Lisoðová, P. Seeman***SM165. Pulsed high-dose dexamethasone versus standard prednisolone treatment for chronic inflammatory demyelinating polyradiculoneuropathy (PREDICT study): a double-blind, randomised, controlled trial.***I.N. van Schaik, F. Eftimov, P.A. van Doorn, E. Brusse, L.H. van den Berg, L. van der Pol, C.G. Faber, J.C.H. van Oostrom, O.J.M. Vogels, R.D.M. Hadden, B.U. Kleine, A.G.W. van Noorden, J.J.G.M. Verschuuren, M.G.W. Dijkgraaf, M. Vermeulen***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. Bouhour, A. Sassolas, C.A. Maurage, F. Couvert*

POSTER SESSION 6C:

PATHOGENIC MECHANISMS OF INHERITED NEUROPATHIES & FRIEDREICH ATAXIA

Chairpersons: R. Baloh, I. Illa

S2

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

A. Roos, M. Baudis, M. Krieger, R.S. Finkel, F. Stanzial, C. Stendel, A. Dufke, H. Topaloglu, K. Zerres, J. Weis, J. Senderek

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 PKC II accumulation in the sciatic nerve following exposure to inflammatory medium

A. Dori, G. Ifergane, T. Saar-Levy, H. Soreq, I. Wirguin

SM175. The first Serbian family with Familial Amyloidotic Polyneuropathy (FAP) (with-drawn)

I. Marjanovic, D. Lavrnica, 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

E. Hutton, L. Carty, M. Laurà, H. Holden, M. Lunn, S. Brandner, R. Mirsky, K. Jessen, M. Reilly

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

T. Kyriakides, E. Dardiotis, G. Hadjigeorgiou, P. Koutsou, K. Christodoulou, M. Saraiva

SM181. The signal transduction pathway of lead exposure induced α -amyloidogenesis and its neuronal damage in human neuroblastoma cell model (withdrawn)

S.S. Chen, C.T. Chen, T.J. Chen

SM182. Disruption of the Cns5/Jab1 gene in Schwann cell alters differentiation and survival causing a peripheral neuropathy

E. Porrello, G. Dina, F. Cerri, D. Triolo, M. Panattoni, U. Del Carro, P. Morana, G. Comi, L. Wrabetz, M.L. Feltri, R. Pardi, A. Quattrini, C.S. Previtali

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. Gourbali, E. Dardiotis, G.M. Hadjigeorgiou, A. Papadimitriou

SM185. Very early electrodiagnostic findings in Guillain-Barré

M.A. Alberti, C. Casanovas, 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. Ajroud-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. Crisculo, 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. Orngreen, P. Laforet, 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, P.A. 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. Aguenouz, 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

G.C. Papadimas, G. Terzis, S. Methenitis, K. Spengos, C. Papadopoulos, S. Kavouras, H. Michelakakis, P. Manta

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, Glycogenesis 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. Itoyama

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

J.B. Lee, J.M. Hong, G.I. Suh, Y.C. Choi, S.J. Na, M. Rajik, P. Kathirvel, W.C. Yee

SM214. TDP-43 pathology in sporadic ALS occurs in motor neurons lacking the RNA editing enzyme ADAR2

H. Aizawa, J. Sawada, T. Hideyama, T. Yamashita, S. Kwak

SM215. Satellite cells dysfunction may contribute to impaired skeletal muscle regeneration in sporadic amyotrophic lateral sclerosis

G. Cenacchi, A. Scaramozza, M. Brini, V. Papa, L. Tarantino, L. Badiali De Giorgi, E. Pegoraro, G. Soraru, C. Angelini

SM216. Lower extremity 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. Atchayaram, 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

M. Aoki, N. Suzuki, H. Warita, M. Kato, H. Mizuno, N. Shimakura, T. Akiyama, H. Furuya, T. Hokonohara, A. Iwaki, S. Togashi, H. Konno, Y. Itoyama

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"

J.L. Muñoz-Blanco, A. Guerrero-Sola, F. Rodríguez de Rivera, J. Esteban, J. Mascías, I. Catalina, L. Galán, C. Buey, J. Mora, E. Díez-Tejedor, J. Matías-Guiu

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

S. Ono, H. Ishikawa, K. Yasui, M. Nomura, H. Mikami, T. Watanabe, M. Suzuki

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)

h. 10.30 – 11.00 PL10. **Diagnosis and treatment of chronic immune mediated neuropathies.**
Eduardo Nobile-Orazio (I)

h. 11.00 – 11.30 PL11. **Clinical Trials in Muscle Diseases: opportunity and challenges.**
Robert C. Griggs (USA)

h. 11.30 – 12.00 PL12. **Spinal muscular atrophy: animal and cellular models to elucidate pathogenesis and design therapeutic approaches**
Judith Melki (France/Israel)

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

h. 14.00 – 15.00 **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

W.K. Engel (Moderator) – M. Dalakas- A. Evoli – E. Nobile-Orazio

h. 15.10 – 17.00 **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 Giacomo, 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

M.C.V. Malicdan, S. Noguchi, Y.K. Hayashi, I. Nonaka, F. Funato, T. Tokutomi, K. Momma, I. Nishino

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. Cwik, 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

L. Peverelli, V. Crugnola, C. Lamperti, V. Lucchini, I. Colombo, M. Ripolone, V. Silani, N. Bresolin, S. Di Mauro, M. Sciacco, G.P. Comi, M. Moggio

S18.PP2. Microarray analysis identifies the gene signature of surviving motor neurones in human SOD1-related 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

S19.L1. Mitochondrial defects in cardiomyopathies.

Giulia D'Amati

S19.L2. Clinical details of Oculopharyngodistal myopathy

Piraye Serdaroglu

S19.L3. Mitochondrial dysfunction in neurodegeneration and aging

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

S19.PP2. Influence of erythrocyte oxygenation and intravascular ATP on the control of skeletal muscle circulation in patients with mitochondrial myopathy

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. Stepniak, 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- γ -sarcoglycan gene therapy for limb girdle muscular dystrophy type 2C

S. Herson, F. Hentati, A. Rigolet, T. Voit, N. Romero, F. Leturcq, P. Lafôret, A. Behin, M. Kefi, M. Audit, M. Montus, C. Masurier, F. Lemoine, F. Carlier, J.Y. Hogrel, H. Haddad, B. Eymard, D. Klatzmann, M. Cherai, D. Caizergues, O. Benveniste

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

N.B.Romero, J.A. Bevilacqua, M. Bitoun, A. Oldfors, F. Lubieniecki, A.L. Taratuto, P. Guicheney, M. Fardeau

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

S. Shalaby, H. Mitsuhashi, C. Matsuda, Y.K. Hayashi, K. Goto, I. Nonaka, S. Noguchi, I. Nishino

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

K.G. Claeys, J.F. Pellissier, F. Garcia-Bragado, J. Weis, A. Urtizberea, J.J. Poza, A.M. Cobo, G. Stoltenburg, D. Figarella-Branger, P.J. Willems, C.E. Depuydt, W. Kleiner, J. Pouget, M.Piraud, G. Brochier, N.B. Romero, M. Fardeau, H.H. Goebel, C.G. Bönnemann, T. Voit, B. Eymard, P. Laforet

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. Salutto, H. Gomez, L. Pirra, A. Dubrovsky, J. Politei, C. Mazia

S22 TW04. Thymolipoma associated with myasthenia gravis

J.H. Cho, S.A. Choi, J.H. Lee, G.S. Kim

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 (TNF α): 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

M. Titulaer, P. Maddison, J. Sont, D. Hilton-Jones, R. Klooster, M. Potman, N. Willcox, P. Sillevs Smitt, A. Vincent, S. van der Maarel, B. Lang, J. Verschuuren

TW11. HLA-DR3 and HLA-B8 are confined to early-onset non-tumour

Lambert-Eaton myasthenic syndrome, similar to autoimmune myasthenia gravis

M. Titulaer, B. Lang, P. Maddison, D. Hilton-Jones, P. Wirtz, A. van der Slik, A. Vincent, B. Roep, N. Willcox, J. Verschuuren

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

E.H. Niks, M.I. Leite, J.B.M. Kuks, M. De Baets, J.G. van Dijk, J. Cossins, D. Beeson, A. Vincent, J. Verschuuren

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

R. Massa, M. Andreani, C. Terracciano, M. Troiano, A. Guagnano, E. Lena, G. Testa, M. Testi

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

T. Masuda, M. Motomura, R. Nakata, M. Tokuda, T. Fukuda, K. Eguchi, T. Yoshimura, M. Tsujihata, K. Utsugisawa

TW18. Clinical and serological features of late-onset myasthenia gravis.

A UK multicentre retrospective cohort study

M.I. Leite, A. Carr, P. Green, B. Mcneillis, P. Maddison, M. Farrugia, J. McConville, D. Hilton-Jones, A. Vincent

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

M. Motomura, R. Nakata, T. Masuda, M. Tokuda, K. Eguchi, T. Yoshimura, H. Shiraishi

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

M.I. Leite, E. Coutinho, P. Waters, A. Graham, E. Santos, L. Jacobson, C. Buckley, D. Hilton-Jones, A. Vincent, J. Palace

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

T. Yoshimura, M. Ito, Y. Honda, T. Fukudome, K. Eric, N. Okada, S. Takeda, M. Motomura, A. Tsujino, T. Masuda, T. Fukuda, S. Yoshimura, K. Ohno

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. Gungor-Tuncer, V. Yilmaz, Y. Parman, P. Serdaroglu, G. Saruhan-Direskeneli, F. Deymeer

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

F.V. Moldovan, M. Negrescu, D. Slavoaca, A. Oancea, A. Stan, A. Blesneag, N. Copil, I. Iuga, Z. Maior, D. Muresanu

TW30. Lambert-Eaton myasthenic syndrome: Clinical and electrophysiological characteristics and response to treatment in 15 Iranian patients

S. Nafissi, R.H.B. Naeeni, M.H. Harirchian, 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

G. Ravenscroft, C. Jackaman, E. McNamara, L. Griffiths, S. Bringans, S. Squire, A. Potter, J. Papadimitriou, K. Davies, N. Laing, K. Nowak

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. Tupler, 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. Khotchenkov, 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. Sblendorio, V. Longo, B. Nico, S. Simonetti, A. De Luca

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

S.K. Mishra, Y. Valles-Ayoub, Z. Khokher, A. Haghghatgoo, D. No, S. Esfandiarifard, C. Saechao, J. Kay, C. Creencia, S. Darvish, D. Darvish

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. Naulet, 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. Sblendorio, 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 mdx^{5Cv} dystrophic mice

O. Dorchies, J. Patte-Reutenauer, O. Patthey-Vuadens, U. Rugg

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. Rugg

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*

F. Saito, A. Nakamura-Okuma, M. Ikeda, H. Hagiwara, T. Shimizu, K. Matsumura

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 $\alpha 5$ and $\alpha 6$ 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. Bellafigliore, 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. Vlodayets, 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

G. Ricci, D. Orsucci, L. Volpi, E. Caldarazzo Ienco, A. LoGerfo, F. Galetta, G. Santoro, A. Rocchi, L. Petrozzi, M. Mancuso, G. Siciliano

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

S. Menazza, B. Blaauw, T. Tiepolo, L. Toniolo, P. Braghetta, A. Pici, B. Spolaore, C. Reggiani, F. Di Lisa, P. Bonaldo, M. Canton

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

E. Perli, C. Giordano, H.L.A. Tuppen, M. Orlandi, C. Autore, A. Pisano, B. Musumeci, R.W. Taylor, M. Montopoli, G. d'Amati

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

C. Quinzii, L.C. Lopez, R. Gilkerson, B. Dorado, J. Coku, A. Naini, C. Lagier-Tourenne, M. Schuelke, L. Salviati, R. Carozzo, 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

C. Quinzii, L.C. Lopez, E. Area, A. Naini, S. Rahman, M. Schuelke, L. Salviati, R. Carozzo, F. Santorelli, S. DiMauro, M. Hirano

TW77. Deleterious mutations in RRM2B, a gene responsible for maintenance of mtDNA, result in severe reduction of mtDNA content in skeletal muscle

R. Van Coster, J. Smet, B. De Paepe, W. Lissens, L. De Meirleir, S. Seneca

TW78. Mutations in TMEM70, a gene coding for an assembly factor of complex V, can cause a severe encephalomyopathy 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, L. De Meirleir, M. Spilioti, X. Fitsioris, A. Evangelidou

TW79. A novel splice site mutation of the POLG gene in two unrelated adults with ophthalmoparesis and myopathy

M. Milone, 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

C. Scuderi, E. Borgione, F. Castello, M. Lo Giudice, R. Pettinato, M. Elia, G.A. Vitello, S.A. Musumeci

TW83. Incidental mitochondrial myopathy: five isolated cases

V. Lucchini, V. Crugnola, L. Napoli, D. Ronchi, M.G. D'Angelo, A. Bordoni, G. Fagiolari, L. Peverelli, N. Bresolin, G. Comi, M. Moggio, M. Sciacco

TW84. MERRF mutation A8344G in a four-generation family without CNS involvement: clinical and molecular characterization

V. Crugnola, L. Peverelli, V. Lucchini, A. Bordoni, P. Ciscato, V. Civelli, D. Spagnoli, N. Bresolin, G. Comi, M. Moggio, M. Sciacco

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

R.D.S. Pitceathly, J.L. Holton, C. Woodward, M.G. Sweeney, C. Fratter, S. Rahman, M.G. Hanna

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. Veggiotti, G.P. Comi

TW90. Partial duplication of mtDNA-tRNA^{Phe} 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 Giacomo, A. Cianfoni, M. Catteruccia, D. Sauchelli, C. Cuccagna, G. Miceli

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

J. Gurgel-Giannetti, M.T. Lara, E.L.C. Concentino, M.C.P. Martins-Machado, L.U. Yamamoto, P. Onofre-Oliveira, M. Vainzof

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 Giorgi, R. Cogliandro, A. Antonucci, L. Iommarini, P. Montagna, R. Liguori, V. Carelli, M.L. Valentino

TW95. Enteropathic arthritis in mitochondrial neurogastrointestinal encephalomyopathy

M. Scarpelli, P. Tonin, M.S. Cotelli, M. Rossi, A. Salvi, V. Vielmi, L. Broglio, M. Tentorio, A. Padovani, G. Tomelleri, M. Filosto

TW96. A novel mutation in the anticodon of the mitochondrial tRNA Arg in a patient with mitochondrial myopathy

A.R. Moslemi, N. Darin, G. Kollberg, M. Tulinius, E. Holme, A. Oldfors

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. Gavrilă

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

Y. Koide, M. Kadoya, M. Hashimoto, A. Kubota, S. Tsuji, J. Shimizu

TW106. Muscle pathology in diseases other than a true myopathies

J.M. Grau, 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, E.F. Hedderick, K.R. Wagner, T.G. Marr, C. Hourde, L.A. Leinwand, D.K. Vaughan, D. Ronald

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

J. Shimizu, M. Hashimoto, M. Kadoya, Y. Koide, A. Kubota, 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**
Mahajan, M.D., Shalini; Engel, M.D., W. King
- TW117. Integrating palliative care across the spectrum of life-threatening pediatric neuromuscular 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. Maués 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.

Wednesday – July 21st, 2010

- h. 8.00 – 10.00 **Poster viewing, coffee and rolls**
- h. 10.00 – 12.00 **PLENARY LECTURES** (*Red and Blue Halls*)
Chairs: Reinhardt Rüdel (Germany) and Byron Kakulas (Australia)
- h. 10.00 – 10.30 **PL13. Development of novel drugs to improve muscle regeneration, and improve muscle function in muscular dystrophy**
Eric Hoffman (USA)
- h. 10.30 – 11.00 **PL14. Therapeutic development in myotonic dystrophy: progress and pitfalls**
Charles Thornton (USA)

- 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**
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***
X. Shen, J. Brengman, G. Acsadi, T. Crawford, S. Iannaccone, E. Karaca, C. Khoury, J. Mah, S. Edvardson, D. Rodgers, A. Engel
- 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. Rakocovic-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

A. Nascimento, G. Alfonso, V. Kata, S. Bernal, L. Alias, C. Ortez, C. Jimenez-Mallebrera, E. Tizzano, J. Colomer

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

L. Baumbach-Reardon

S24 (Blue Hall)

Exercise 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. Kiliç, S. Aksu Yıldırym, S. Atay Yılmaz, 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. Thøgersen, N. Preisler, M. Berthelsen, J. Vissing

S25 (Hall E2)

Friedreich's Ataxia

Chairs: 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

Chairs: Vincenzo Nigro and Diane Shelton

Invited Speakers

S26.L1. Systemic delivery of genes to striated muscles: potential for therapy of muscular dystrophy

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

K. Charton, N. Daniele, A. Vihola, C. Roudaut, F. Monjaret, A. Tarrade, J. Sarparanta, B.Udd, I. Richard

S26.PP2. Etanercept suppressed the necrosis and inflammatory change in muscle of SJL/J mice by blocking TNF α function

H. Nemoto, S. Konno, H. Sugimoto, N. Nomoto, H. Nakazora, M. Murata, T. Toda, H. Kitazono, T. Fujioka

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

S27.PP1. A Cooperative International Neuromuscular Research Group study of the relationship between impairment, activity limitation, participation and quality of life in persons with confirmed dystrophinopathies: One year follow-up of skeletal muscle strength and timed motor performance.

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

S28.L3. Skeletal muscle channelopathies

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. Sblendorio, 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. 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

S20

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. Palmiter, J. Chamberlain

TW122. Codon-optimized AAV vector for gene therapy in BIO14.6 hamsters

I.L. Rotundo, A. Lancioni, Ge. 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

N. Vieira, C.R. Bueno Junior, V. Brandalise, H.V.A. Caetano, E. Zucconi, M. Secco, M. Vainzof, M. Zatz

TW124. Lesson from the patients skipped by nature: characterization of Becker Muscular Dystrophy (BMD) with Exon 51 as one endpoint

S. Cirak, V. Arechavala-Gomez, 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

L. Previato de Almeida, J.E. Cury Fiuza, A.F. Gembre, T. Malardo, I. Cardoso Fontoura, R. Ariza Rosseti, J. C. S. Bizário, C. Lopes Silva, A. A.M. Coelho-Castelo

TW128. Wound-healing MRL-MpJ phenotype improves outcome of dystrophin-deficient *mdx* mice

Y. Sunada, Y. Ohsawa, M. Fujino, T. Okada, S. Hayashi, M. Rikimaru, T. Murakami, S. Nishimatsu, T. Nohno, M. Nagao

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

T. Domi, E. Porrello, A. Capotondo, D. Triolo, M. Sampaolesi, S. Brunelli, G. Comi, M.A. Ruegg, T. Shin'Ichi, G. Cossu, A. Biffi, A. Quattrini, S.C. Previtali

TW130. The bone marrow mesenchymal stem cells can rescue damaged skeletal muscle

A.B. Liu, C.Y. Pang, C.H. Wang

S27

TW131. Outcome measures validation study for mesoangioblasts transplantation in children affected by Duchenne Muscular Dystrophy

S. Bonfiglio, A. Lerario, S. Markt, S. Napolitano, S. Previtali, M. Scarlato, M.G. Natali-Sora, N. Bresolin, G. Comi, R. Gatti, F. Ciceri, G. Cossu, Y. Torrente, AIM group for DMD

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

R. Barohn, J. Statland, Y. Wang, R. Richesson, B. Bundy, L. Herbelin, J. Gomes, J. Trivedi, S. Venance, A. Amato, M. Hanna, R. Griggs, CINCH Consortium

TW135. Twelve-month interim results of an open label extensions study (DELPHI-E) with idebenone (Catena®) in Duchenne Muscular Dystrophy (DMD)

G.M. Buysse, 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 its causes and the treatment outcomes

S. Misevic, B. Kovac, D. Vukasinovic-Soljagic, 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

S27

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. Reyl

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)

R. Arakawa, K. Yabuuchi, M. Takemaru, T. Okazaki, Y. Hazama, T. Hanaoka, T. Kumamoto

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. Rakocevic-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

R. Petty, C. Longman, M. Di Marco, R. McWilliam, D. Wilcox, K. Naismith

TW155. New challenges in the management of neuromuscular disorders:

the need for a “medicine” of neuromuscular disorders

K. Gorni, P. Banfi, E. Roma, N. Cellotto, C. Lunetta, M. Corbo for NEMO Group

TW156. Reproducibility and reliability of quantitative MRI measures

for neuromuscular disease

J.M. Morrow, C.D.J. Sinclair, M.M. Reilly, M.G. Hanna, T.A. Yousry,

X. Golay, J.S. Thornton

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?

C.D.J. Sinclair, R.S. Samson, D.L. Thomas, N. Weiskopf, A. Lutti, J.S. Thornton, X. Golay

POSTER SESSION 16:

CONGENITAL MYOPATHIES

Chairpersons: C. Wallgren-Pettersson, N. Romero

S21

TW159. Atypical or unfrequent muscular disorders in neuropaediatric syndromes

G.T. Serratrice, B. Chabrol

TW160. Analysis of the nebulin gene by multiplex-ligation 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. Tajsharghi, 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)

M. von der Hagen, W. Kress, A. Ferreira, C. Bönnemann, U. Grieben, C.G. Korenke, U. Reuner, G. Schreiber, M. Stoetter, P. Richard, E. Willichowski, J. Luetsch, U. Schara

TW163. New data on a case of congenital fibre type disproportion and non compaction cardiomyopathy 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. Kumada, 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

C. Ortez, A. Nascimento, C. Jimenez-Mallebrera, A. Gutierrez, C. Jou, J. Corbera, N. Monnier, J. Lunardi, M. Olive, I. Ferrer, J. Colomer

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

E. Yiu, A. Klausegger, L. Waddell, L. Lloyd, N. Grasern, K. Tran, K. North, J. Bauer, C.W. Chow, D. Murrell, M. Ryan

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. Nyamkishig

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

X. Stalpers, A. Verrips, A. Mostert, J. Braakhekke, A. Van den Wijngaard, M. Lammens

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 fibrates in skeletal muscle: a proteomic and genomic approach

G.M. Camerino, S. Pierno, C. Digennaro, 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

F.J. Authier, M. Couette, E. Passeri, P. Maison, P. Brugieres, R. Gherardi, A.C. Bachoud-Levi

TW180. MuRF-1 gene deletion prevents atrophy of disused muscle in the hindlimb-unloaded mice

J.F. Desaphy, S. Pierno, C. Digennaro, 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. Fenicia, F. Anniballi, U. Sinardi, C. Rodolico, A. Toscano

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

I. Gáti, O. Danielsson, G. Leijon, K. Janiec, L. Brattström, B.A. Fredriksson, M. Vrethem, H. Lindehammar, N. Dizdar

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. Guvenc, 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

P. Bernasconi, C. Cappelletti, P. Cavalcante, L. Colleoni, E. Coccia, M. Severa, F. Baggi, F. Zolezzi, G. Tuana, P. Confalonieri, L. Morandi, F. Blasevich, R. Mantegazza

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

S. Sacconi, L. Feasson, J.C. Antoine, C. Pecheux, R. Bernard, A.M. Coubo, A. Casarin, L. Salviati, C. Desnuelle, A. Urtizberea

TW187. Evaluation of two different methods for per-oral gastrostomy tube placement in patients with motor neuron disease (MND): PIG versus PEG procedures

C. Mc Dermott, 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

A.S.D. Saporta, S.L. Sottile, S.M.E. Feely, L.J. Miller, C.E. Siskind, M.E. Shy

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

S. Cagnin, S. De Palma, P. Martini, C. Borsato, M. Fanin, R. Salarioli, M. Vasso, T. Raffaello, G. Cenacchi, C. Angelini, C. Gelfi, G. Lanfranchi

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 31st, 2010: € 600

Payment received after March 31st, 2010: € 750

Persons under 35 years of age:

Payment received by March 31st, 2010: € 320

Payment received after March 31st, 2010: € 700

Accompanying persons:

Payment received by March 31st, 2010: € 220

Payment received after March 31st, 2010: € 220

WANDA delegates:

Payment received by March 31st, 2010: € 320

Payment received after March 31st, 2010: € 700

One-day registration (Wednesday 21st July): € 100

Teaching courses participants:

Payment received by March 31st, 2010: € 100

Payment received after March 31st, 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 1st, 2010: Submission of Abstracts

May 31st, 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 Environs

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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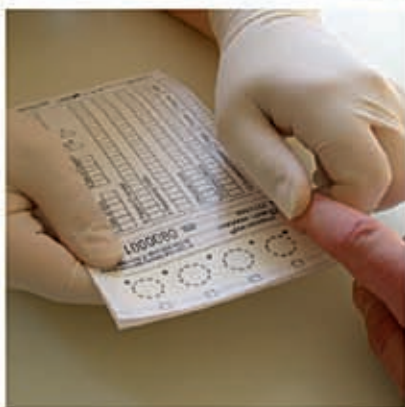
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1. Mellias, U. and Lofaso, F. Pompe disease: A neuromuscular disease with respiratory muscle involvement. *Respir Med*, 2009; 103: 477-84.
2. Kidman, P., Steiner, R., Ball, D., et al. Pompe disease diagnosis and management guideline. *Chest Med*, 2006; 6: 267-288.
3. Kallwies, H., Carr, C., Germain, J., et al. Rapid diagnosis of late-onset Pompe disease by fluorimetric assay of alpha-glucosidase activities in dried blood spots. *Mol Genet Metab* 2007; 90: 449-52.

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Confirmatory diagnosis can be made
quickly and accurately in blood
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1. Kikland JS et al. Pompe disease diagnosis and management guideline. *Genetics in Medicine*, May 2006, Vol. 8 No. 5, page 267-288
2. Kallunki H et al. Rapid diagnosis of late-onset Pompe disease by fluorometric assay of α -glucosidase activities in dried blood spots. *Molecular Genetics Metabolism* 90, page 449-452
3. Chinnels NA et al. Glycogen storage disease type II: enzymatic screening in dried blood spots on filter paper. *Clin Chim Acta* 2004; 347:97-102
4. Jai K et al. The use of acarbose inhibition in the measurement of acid α -glucosidase activity in blood lymphocytes for the diagnosis of Pompe disease. *Genet Med* 2006; 8:107-112.



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