

CONGRESSO CONGIUNTO AIM/ASNP
Digital Edition
9-12 Dicembre 2020

PROGRAMME

Mercoledì 9 Dicembre Streaming on CHANNEL 1

14.00 – 14.20 | Greetings and introduction

14.20 - 15.20 Workshop:
Innovative therapeutic strategies in genetic neuromuscular diseases (Part I)

14.20 – 14.40 An “antisense” approach for neuromuscular diseases | *Toby Ferguson*

14.40 - 15.00 Gene therapy for SMA: from an innovative mechanism of action to clinical data | *M. Pane*

15.00 – 15.20 In vivo gene therapy for Pompe disease | *F. Mingozzi*

15.20 - 16.40 | Workshop:
New tools in the management of patients with neuromuscular disorders

15.20 - 15.40 Genetic modifiers as a tool for personalized treatment of neuromuscular disorders

L. Bello

15.40 – 16.00 Animal models as a tool to design novel therapeutical strategies | *S. Previtali*

16.00 – 16.20 Computation Neurology: devices, data and models for characterization and modelling of neuromuscular diseases | *V. Sanguineti*

16.20 – 16.40 Digital health solutions to increase sensitivity of outcome measures in clinical and trial settings in neuromuscular disorders | *S. Messina*

16.40 - 16.50 | Pausa

16.50 - 18.20 | Oral communications 1: “Updating diagnostic tools in neuromuscular disorders”

16.50 – 17.05 Automatic recognition of ragged red fibers in muscle's histological images of patients affected by mitochondrial disorders

J. Baldacci, M. Calderisi, A. Rubegni, C. Fiorillo, F.M. Santorelli (Pisa; Genova)

17.05 – 17.20 Ultra-high frequency ultrasound of median nerve. Comparison of different frequencies and correlation with clinical and neurophysiological findings

D. Coraci, A. De Luna, A. Fusco, S. Giovannini, L. Padua (Roma; Milano)

17.20 – 17.35 Muscle MRI in Myotonic Dystrophy type1 (DM1) *M. Garibaldi, T. Nicoletti, L. Fionda, E. Bucci, T. Tartaglione, G. Tasca, A. Perna, A. Petrucci, G. Silvestri, G. Antonini (Roma)*

17.35 – 17.50 Virtual genetic counselling in Neuromuscular diseases: a pilot telegenetics project
F. Gualandi, M. Neri, M. Farnè, F. Fortunato, G. Vita, M. Pane, G. Rocchi, T. Evangelista, A. Ferlini (Ferrara; Messina; Roma; Pesaro; Paris, France)

17.50 – 18.05 Whole exome and genome sequencing for the genetic diagnosis of dystrophinopathies
R. Selvatici, R. Rossi, F. Mingyan, M.S. Falzarano, P. Rimessi, M. Neri, F. Gualandi, S. Delin, S. Bensemmane, A. Shatillo, L. Bello, E. Pegoraro, A. Ferlini (Ferrara; Shenzhen, China; Zadar, Croatia; Alger; Kharkiv, Ukraine; Padova)

18.05 – 18.20 Skin biopsy may help to distinguish Multiple System Atrophy-parkinsonism type from Parkinson disease with orthostatic hypotension
V. Donadio, A. Incensi, G. Rizzo, R. De Micco, A. Tessitore, G. Devigili, F. Del Sorbo, S. Bonvegna, M. Magnani, C. Zenesini, L. Vignatelli, R. Cilia, R. Eleopra, G. Tedeschi, R. Liguori (Bologna; Napoli; Milano)

18.20 – 18.50 | Lecture:

Neuromuscular disorders prevention: newborn screening, prenatal and pre-implantation diagnosis and counseling | *L. Stuppia*

18.50 – 20.10 | Workshop:

Innovative diagnostic aspects in neuromuscular disorders

18.50 – 19.10 New histopathological markers for the diagnosis of the myopathies: the vintage that is updated | *C. Fiorillo*

19.10 – 19.30 Skin biopsy: beyond the intraepidermal nerve fiber (IENF) density | *M. Nolano*

19.30 – 19.50 Genome analyses in muscular disorders | *V. Nigro*

19.50 – 20.10 NGS in CMT-related disorders | *F. Taroni*

Giovedì 10 Dicembre
Streaming on CHANNEL 1

08.30 – 9.30 | Breakfast Seminar:
Transthyretin-related Amyloidosis

Chairmen:

8.30 – 8.50 - Molecular mechanisms | L. *Obici*

8.50 – 9.10 Clinical presentation, diagnosis and treatment of peripheral neuropathy | G. *Vita*

9.10 – 9.30 Clinical and therapeutic aspects of cardiomyopathy | G. *Limongelli*

9.30 - 11.30 | Oral communications 2: “Inherited neuromuscular disorders”

9.30 - 9.45 Clinical, morphological and genetic data of patients with distal and myofibrillar myopathies: report from the Italian network.

S. Bortolani, S. Bonanno, G. Vattei, P. Tonin, M. Monforte, E. Ricci, G. Primiano, S. Servidei, G. Greco, R. Massa, C. Gemelli, M. Grandis, C. Fiorillo, A. Petrucci, M. Filosto, M.L. Valentino, R. Liguori, T. Mongini, M. Garibaldi, G. Antonini, M. Lucchini, M. Mirabella, A. Rubegni, F. M. Santorelli, G. Siciliano, G. Ricci, C. Angelini, A. Ariatti, L. Maggi, G. Tasca (Roma; Verona; Milano; Genova; Brescia; Bologna; Torino; Pisa; Venezia; Modena)

9.45 - 10.00 Phenotypic and genetic characterization of childhood Charcot-Marie-Tooth disease

F.R. Danti, S. Magri, S. Genitrini, E. Pagliano, M. Foscan, A. Ardisson, C. Ciano, P. Saveri, F. Balistreri, D. Di Bella, D. Pareyson, F. Taroni, I. Moroni (Milano)

10.00 - 10.15 Mutations in Supervillin cause myopathy with myofibrillar disorganization and autophagic vacuoles

C. Hedberg-Oldfors, R. Meyer, K. Nolte, Y. Abdul Rahim, C. Lindberg, K. Karason, I. J. Thuestad, K. Visuttijai, M. Geijer, M. Begemann, F. Kraft, E. Lausberg, L. Hitpaß, R. Götzl, E.J. Luna, H. Lochmüller, S. Koschmieder, M. Gramlich, B. Gess, M. Elbracht, J. Weis, I. Kurth, A. Oldfors, C. Knopp (Gothenburg, Sweden; Aachen, Germany; Malmö, Sweden; Worcester, United States; Ottawa, Canada)

10.15 - 10.30 Clinical and biological characterization of a large series of late-onset CMT21 patients carrying the MPZ P70S mutation

P. Saveri, C. Pisciotta, M. Grandis, V. Prada, P. Fossa, R. Mastrangelo, C. Ferri, G. Shackelford, F. Veneri, R. Baldi, G. Lauria, R. Lombardi, C. Ciano, S. Magri, F. Taroni, L. Richard, J.M. Vallat, M. D’Antonio, D. Pareyson (Milano; Genova; Limoges Cedex, France)

10.30 - 10.45 Long-term follow-up and clinical features in an Italian cohort of patients with GNE myopathy.

A. Pugliese, C. Bonanno, G. Nicocia, A. Lupica, S. Messina, GL. Vita, G. Vita, A. Toscano, C. Rodolico (Messina)

10.45 - 11.00 Peripheral nerve involvement in VCP-related multisystem proteinopathy.

S. Testi, M. Filosto, A. Mazzeo, M. Sabatelli, M. Luigetti, E. Pancheri, G. Vattermi, P. Tonin, T. Cavallaro, G.M. Fabrizi (Verona; Brescia; Messina; Roma)

11.00 - 11.15 Congenital Myasthenic Syndromes: a large Italian cohort of patients

A. Gallone, R. Brugnoli, A. Ardisson, E. Terlizzi, R. Masson, G. Ricci, F. Magri, F. Guidolin, M.L. Valentino, D. Frattini, C. Bonanno, M. Catteruccia, A. Malandrini, G. Primiano, C. Antozzi, P. Confalonieri, G. Tasca, M. Monforte, E. Ricci, G. Astrea, C. Ticci, M. Garibaldi, D. Orsucci, F. Ricci, M.T. Ferrò, V.A. Donadio, A. Gentili, E. Bertini, G. Siciliano, D. Piga, G.P. Comi, R. Liguori, I. Moroni, F.M. Santorelli, S. Servidei, A. D'Amico, G. Antonini, A. Evoli, D. Sternberg, D. Beeson, A. Engel, H. Lochmüller, R. Mantegazza, P. Bernasconi, C. Rodolico, L. Maggi (Milano; Piacenza; Pisa; Trieste; Bologna; Reggio Emilia; Messina; Roma; Siena; Lucca; Torino; Crema; Paris, France; Oxford, UK; Rochester, MN, USA; Freiburg, Germany; Ottawa, Canada)

11.15 - 11.30 Preliminary results of rank approach for improving quality of life in Charcot-Marie-Tooth type 1A patients

S. Tozza, D. Severi, D. Bruzzese, R. Iodice, L. Ruggiero, R. Dubbioso, E. Spina, A. Iovino, F. Aruta, M. Bellofatto, M. Nolano, L. Santoro, F. Manganelli (Napoli)

11.30 - 12.00 | Lecture: “High-cost drugs economic sustainability for the National Health Service. How long can we afford them?” | R. Tarricone

12.00 – 12.15 | Pausa

12.15 -13.15 | Workshop:

Imaging techniques in muscle and peripheral nerve diseases

12.15- 12.35 Ultrasound as a multi-tool for Muscle and Nerve | *L. Padua*

12.35 -12.55 MRI in peripheral nerve disorders | *S. Gerevini*

12.55 - 13.15 Imaging vs morphology in the current diagnostic workup of muscle disorders
M. Garibaldi

13.15 – 14.00 – POSTER SESSION (Streaming on Channel 2, 3, 4, 5 ,6 ,7 ,8, 9)

13.15 – 14.00 - CHANNEL 2 – Updating diagnostic tools in neuromuscular disorders

13.15 – 13.20 Muscle MRI pattern recognition: a sample study using a MRI-based tool

M. Maffei, M. Giannotta, G. Scarpini, L. Cirignotta, A. Pini (Bologna)

13.20 – 13.25 Guillain Barrè syndrome chameleon: gadolinium magnetic resonance helps in early diagnosis

Salvalaggio, F. Castellani, M. Anglani, M. Campagnolo, R. Manara, C. Briani (Padova)

13.25 – 13.30 Six muscular MRI patterns in search for an author: clinical, genetic and imaging characterization of six myopathic/dystrophic case reports without a definite diagnosis

M. Rossi, M. Paoletti, V. Vacchini, A. Ferrero, S. Parravicini, A. Gardani, A. Asaro, A. Pichiecchio, A. Berardinelli (Pavia)

13.30 – 13.35 Genilam, an Italian Project to shorten the time of molecular diagnosis for ATTR amyloidosis patients

D. Bonvissuto, E. Rizzo, F. Franchini, A. Nuccitelli, A. Biricik, C. Grillo, L. Barbetta, F. Fiorentino (Roma)

13.35 – 13.40 Unusual findings detected by diagnostic gene panel sequencing applied to heterogenous neuromuscular disorders

F. Magri, G. Manenti, R. Brusa, P. Ciscato, R. Del Bo, F. Fortunato, S. Lucchiari, S. Pagliarani, D. Piga, D. Velardo, V. Sansone, T. Mongini, S. Gandossini, S. Corti, M. Moggio, G.P.Comi, D. Ronchi (Milano; Lecco; Torino; Bosisio Parini)

13.40 – 13.45 NEUROMIO a custom NGS based panel for neuromuscular disorders diagnosis: results of the analysis in a cohort of patients

M. Neri, F. Fortunato, C. Trabanelli, P. Rimessi, R. Selvatici, D. Ognibene, S. Bigoni, S. Fini, E. Terlizzi, D. Giachino, ML Valentino, LM Rocchetti, I. Donati, V. Uliana, E. Pegoraro, A. Pini, M. Pane, F. Gualandi, A. Ferlini (Ferrara; Piacenza; Torino, Orbassano; Bologna; Cesena; Parma; Padova; Roma)

13.45 – 13.50 Whole genome sequencing in a pair of siblings affected with Duchenne muscular dystrophy with discordant cognitive phenotype

D. Sabbatini, M. Alexander, S. Vianello, A. Fusto, B. Merlo, M. Villa, V. Zangaro, F. De Pascale, G. Sorarù, E. Pegoraro, L. Bello (Padova; Birmingham AL, U.S.A.; Padova)

13.50 – 13.55 Promoting Early Diagnoses in Neuromuscular disease (PEDINE). A pilot study

M. Vacchetti, C. Brusa, R. D'Alessandro, M. Bobbio, M. Spada, R. Turra, F. Ricci, T. Mongini, PEDINE working group (Torino)

13.55 – 14.00 Abnormal α -synuclein deposits in skin nerves: inter and intra-laboratory reproducibility

V. Donadio, K. Doppler, A. Incensi, A. Kuzkina, A. Janzen, J. Volkmann, G. Rizzo, E. Antelmi, G. Plazzi, C. Sommer, WH Oertel, R. Liguori (Bologna; Marburg, Germany)

13.15 – 14.00 CHANNEL 3 - Inherited neuromuscular disorders

13.15 – 13.20 A genotyping and clinical neurophysiological study of early-onset Charcot-Marie-Tooth disease

C. Croci, P. Mandich, I. Meola, A. Geroldi, E. Bellone, A. Accogli, C. Bruno, M. Pedemonte, C. Minetti, C. Fiorillo, P. Lanteri (Genova; Alessandria; Milano)

13.20 – 13.25 Pilot study in phenotypic variability of cardiac involvement in a cohort of patients with Becker muscular dystrophy

V. Castiglione, G. Ricci, A. Govoni, G. Astrea, A. Rocchi, F. Baldinotti, A. Giannoni, C. Passino, M. Emdin, G. Siciliano (Pisa)

13.25 – 13.30 Clinical and genetic characteristics of NEFL-related Charcot – Marie Tooth disease due to P440L mutation in a large Italian family

A. Petrucci, L. Lispi, M. Garibaldi, E. Frezza, R. Massa, F. Moro, F.M. Santorelli (Roma; Pisa)

13.30 – 13.35 Expanding the phenotype of p.R1460W mutation in SCN4A gene: a family report

S. Cotti Piccinelli, B. Risi, E. Baldelli, N. Necchini, A. Galvagni, A. Padovani, R. Brugnoli, L. Maggi, M. Filosto (Brescia; Milano)

13.35 – 13.40 Charcot – Marie – Toths disease and pregnancy: data from the Italian CMT national registry

C. Pisciotta, D. Calabrese, L. Santoro, I. Tramacere, F. Manganelli, G.M. Fabrizi, A. Schenone, T. Cavallaro, M. Grandis, S. Previtali, I. Allegri, L. Padua, C. Pazzaglia, A. Quattrone, P. Valentino, S. Tozza, A. Mazzeo, M.C. Trapasso, F. Parazzini, G. Vita, D. Pareyson; for the Italian CMT Network (Milano; Napoli; Verona; Genova; Parma; Catanzaro; Messina)

13.40 – 13.45 Alpha-sarcoglycanopathy presenting as myalgia and hyperckemia in two adults with a long-term follow-up

C. Dosi, A. Rubegni, D. Cassandrini, A. Malandrini, L. Maggi, F.M. Santorelli (Pisa; Siena; Milano)

13.45 – 13.50 A new Italian family with PMP2-related Charcot-Marie-Tooth Disease type 1G

S. Spolverato, F. Taioli, G. Zanette, S. Romito, M. Ferrarini, L. Roncari, G. Cantalupo, G.M. Fabrizi (Verona; Pescheria del Garda)

13.50 – 13.55 Exon 1 nonsense mutation of dystrophin gene and exception to the reading-frame rule

A. Govoni, G. Ricci, A. Lo Gerfo, V. Castiglione, L. Calì, F. Baldinotti, A. Rocchi, M. Emdin, G. Siciliano (Pisa)

13.55 – 14.00 Severe Charcot-Marie-Tooth disease type 1A in a patient with PMP22 tetrasomy

F. Taioli, G.P. Zanette, M. Ferrarini, S. Testi, T. Cavallaro, G.M. Fabrizi (Verona; Peschiera del Garda)

13.15 – 14.00 - CHANNEL 4 - Inherited neuromuscular disorders

13.15 – 13.20 Clinical presentations of two rare mutations of TTR

D. Cardellini, F. Taioli, M. Cappellari, M. Milan, L. Bertolasi, G.M. Fabrizi, T. Cavallaro (Verona; Venezia)

13.20 – 13.25 Mutations in ASCC3 are associated with severe congenital myopathy, arthrogyriposis and bone fractures *M. Catteruccia, D. Diodato, F. Fattori, G. Colia, F. De Mitri, A. D'Amico, E. Bertini (Roma)*

13.25 – 13.30 Sensorimotor axonal polyneuropathy and a VUS mutation in Transthyretin gene *F. Gragnani, F. Gilio, R. Iulianella, I.F. Pestalozza, F. Cortese, A.M. Cipriani, E.M. Pennisi (Roma)*

13.30 – 13.35 A de novo in-frame deletion in MYOT causes an early adult onset severe distal myopathy *E. Pancheri, V. Guglielmi, V. Nigro, S. Aurino, A. Torella, M. Malatesta, A. Vettore, A. Giorgetti, G. Tomelleri, P. Tonin, G. Vattei (Verona; Napoli)*

13.35 – 13.40 Description of the first cohort of V122I ATTRv amyloidosis patients from non-endemic areas *M. Russo, L. Gentile, G. Di Bella, F. Minutoli, F. Cucinotta, L. Obici, R. Mussinelli, A. Toscano, G. Vita, A. Mazzeo (Messina; Pavia)*

13.40 – 13.45 Whole Exome Sequencing identifies two novel candidate genes and extends the diagnostic spectrum of patients with neuromuscular diseases *R. Rossi, M.S. Falzarano, M. Pinotti, D. Balestra, M. Neri, F. Fortunato, E. Mercuri, M. Pane, F. Gualandi, R. Selvatici, A. Ferlini (Ferrara; Roma)*

13.45 – 13.50 Hereditary Transthyretin Amyloidosis (hATTR) Polyneuropathy: experience from a single center *D. Severi, S. Tozza, E. Spina, M. Bellofatto, M. Nolano, L. Santoro, F. Manganelli (Napoli)*

13.50 – 13.55 Tubular aggregate myopathy: clinical and molecular characterization in STIM1 and ORAI1 mutated patients *S. Tripodi, L. Bello, G. Minervini, C. Reggiani, S. Vianello, V. Bozzoni, L. Caumo, P. Riguzzi, A. Lupi, C. Bertolin, G. Sorarù, E. Pegoraro (Padova)*

13.55 – 14.00 Clinical features in ATTRv patients *S. Tozza, D. Severi, P. Origone, A. Geroldi, E. Bellone, P. Mandich, M. Nolano, L. Santoro, F. Manganelli (Napoli; Genova)*

13.15 – 14.00 CHANNEL 5 - Inherited neuromuscular disorders

13.15 – 13.20 Recommendations for presymptomatic genetic testing for ATTRv in the era of effective therapy: a multicenter Italian study *M. Grandis, L. Obici, G. Ferrandes, D. Pareyson, C. Briani, C. Danesino, M. Canepa, A. Mazzeo, L. Trevisan, S. Fenu, F. Cappelli, F. Perfetto, L. G. Pradotto, F. Benedicenti, G.M. Fabrizi, G. Bisogni, M. Luigetti, P. Rimessi, L. Melchiorri, G. Tini, C. Gemelli, S. Tozza, D. Severi, P. Mandich (Genova; Pavia; Milano; Padova; Messina; Firenze; Oggebbio (Verbania); Bolzano; Verona; Roma; Ferrara; Napoli)*

13.20 – 13.25 Molecular and clinical characterization in asymptomatic and symptomatic DMD carriers

V. Zangaro, P. Riguzzi, B. Merlo, S. Vianello, R. Bariani, L. Lamagna, B. Bauce, G. Sorarù, L. Bello, E. Pegoraro (Padova)

13.25 – 13.30 Hereditary neuropathy with liability to pressure palsy: two pediatric symptomatic cases with acute monolateral foot drop

G. Scarpini, M. Giannotta, F. Pastorelli, A. Pini (Bologna)

13.30 – 13.35 UNC-45B mutations in congenital axial myopathy with cores and multiple internalised nuclei

C. Fiorillo, C. Panicucci, S. Donkervoort, M. Traverso, P. Broda, R. Falsaperla, V. Salpietro, C. Minetti (Genova)

13.35 – 13.40 A case of a Heterozygous Kv1.1 N255D Mutation with Normal Serum Magnesium Levels

F. Bianchi, C. Simoncini, G. Ricci, P. Bernasconi, G. Siciliano (Pisa; Milano)

13.40 – 13.45 Clinical and electrophysiological features of seipinopathy in an Italian family: an example of spontaneous regressive course of the disease - a case report

S. Parravicini, A. Asaro, C. Cereda, A. Lozza, A. Berardinelli (Pavia)

13.45 – 13.50 Familial Progressive Cardiac Conduction Disease caused by a TRPM4 gene mutation

A. Palladino, Torella, R. Petillo, A. M. Scutifero, S. Morra, P. D'Ambrosio, L. Passamano, V. Nigro, L. Politano (Napoli)

13.50 – 13.55 Combined genetic causes of rhabdomyolysis in a family: can it be a double trouble?

F. Torri, G. Ricci, C. Simoncini, P. Annoscia, G. Alì, L. Chico, D. Cassandrini, Di Muzio, G. Siciliano (Pisa; Chieti)

13.55 – 14.00 Myotonic dystrophy type 1: daytime pulmonary function and sleep related breathing disorders

S. De Pasqua, F. Pizza, P. Avoni C. Quarta, G. Plazzi, R. Liguori (Bologna)

13.15 – 13.50 - CHANNEL 6 - Biomarkers and outcome measures in neuromuscular disorders

13.15 – 13.20 Fatigue and sleepiness in myotonic dystrophy type 1: motor, neuropsychological and sleep correlates

E. Frezza, C. Galluzzi, G. Greco, P. Proserpio, L. Mauro, G. Sannicolò, A. Pirola, E. Falcier, E. Roma, A. Zanolini, A. Lizio C. Liguori, F. Placidi, V.A. Sansone, R. Massa (Roma; Milano)

13.20 – 13.25 A possible biomarker for paclitaxel-induced peripheral neurotoxicity: Neurofilament light chain serum levels monitoring

P. Alberti, F. Cicchiello, F. Riva, G. Cavaletti, M.E. Cazzaniga (Monza)

13.25 – 13.30 Gender effect on cardiac involvement in Myotonic Dystrophy type 1 (DM1)

M. Garibaldi, L. Fionda, E. Bucci, F. Vanoli, L. Leonardi, A. Lauletta, G. Alfieri, G. Merlonghi, S. Morino, M. Testa, G. Antonini (Roma)

13.30 – 13.35 Serum neurofilament light chain (sNfL) correlate with sural nerve pathological findings

F. Castellani, S. Mariotto, M. Campagnolo, S. Ferrari, C. Briani (Padova; Verona)

13.35 – 13.40 Cognitive and personality involvement in DM1 patients: is there an age at onset related effect?

E. Lai, G. Spadoni, C. Simoncini, G. Ricci, G. Siciliano (Pisa)

13.40 – 13.45 Definition of a new ICF core-set for the upper limbs of hereditary neuropathies

V. Prada, B. Mazzarino, A. Mazzeo, D. Pareyson, L. Santoro, L. Padua, G. Fabrizi, A. Schenone and ULNA group (I. Poggi, L. Mori, M. Grandis, C. Gemelli, L. Gentile, A. Tisano, F. Cavallaro, C. Pisciotto, M. Montesano, F. Manganelli, S. Tozza, G. Aceto, D. Dellaventura, C. Pazzaglia, C. Erra, D. Coraci, T. Cavallaro, A. Peretti, L. Roncari) (Genova; Messina; Milano; Napoli; Roma; Verona)

13.45 – 13.50 Myotonic Dystrophy type 2 unmasked by physical exercise following COVID-19 lockdown

S. Lucchiari, F. Magri, G.P. Comi, M. Sciacco (Milano)

13.15 – 13.50 CHANNEL 7 - Biomarkers and outcome measures in neuromuscular disorders

13.15 – 13.20 Gait and balance evaluation in patients with different myopathies: correlation between imaging data and functional aspects

A. Modenese, A. Picelli, N. Mattiuz, E. Pancheri, N. Smania, G. Vattermi, P. Tonin (Verona)

13.20 – 13.25 Issues in the management of hereditary transthyretin-mediated amyloidosis (hATTR)

A. Parachini, B. Valzasina, M. Grandis, M. Oliverio, A. Schenone, L. Obici (Pavia)

13.25 – 13.30 Capturing the patient-reported impact of myasthenia gravis in the real-world setting using a smartphone application

F. Saccà, S. Berrih-Aknin, K.G. Claeys, R. Mantegazza, H. Murai, E. Bagshaw, H. Kousoulakou, M. Larkin, T. Leighton, S. Paci (Napoli; Paris, France; Leuven, Belgium; Milano; Narita, Japan; Oxford, United Kingdom; Ghent, Belgium)

13.30 – 13.35 Spinal cord volumetry as a biomarker for monitoring drug treatment in spinal muscular atrophy patients: a pilot MRI study

G. Savini, C. Asteggiano, L.M. Farina, C.A.M. Gandini Wheeler-Kingshott, S. Bastianello, A. Berardinelli, A. Pichiecchio (Pavia; London, UK)

13.35 – 13.40 Muscle MRI of upper girdle in GNE myopathy

E. Torchia, M. Lucchini, S. Bortolani, M. Monforte, M. Mirabella, E. Ricci, G. Tasca (Roma)

13.40 – 13.45 Real-World Treatment Patterns and Outcomes in Patients With Spinal Muscular Atrophy Collected From the RESTORE Registry

L. Servais, J.W. Day, D.C. De Vivo, J. Kirschner, E. Mercuri, F. Muntoni, P.B. Shieh, E. Tizzano, I. Desguerre, S. Quijano-Roy, K. Saito, M. Droege, O. Dabbous, R. Cerbini, F. Balducci, A. Shah, F. Khan, F.A. Anderson, R.S. Finkel (Oxford, United Kingdom; Stanford, CA, United States; New York,

NY, United States; Freiburg, Germany; Roma; London, United Kingdom; Los Angeles, CA, United States; Barcelona, Spain; Paris, France; Garches, France; Tokyo, Japan; Bannockburn, IL, United States; Worcester, MA, United States; Orlando, FL, United States)

13.45 – 13.50 Hind limb unloading as a model of skeletal muscle atrophy: validation of in vivo and ex vivo readouts for preclinical translational research

P. Mantuano, F. Sanarica, O. Cappellari, B. Boccanegra, N. Tarantino, E. Conte, M. De Bellis, G. M. Camerino, S. Pierno, A. De Luca (Bari)

13.15 – 13.50 CHANNEL 8 - Biomarkers and outcome measures in neuromuscular disorders

13.15 – 13.20 HyperCKemia and viral infections: a lesson from COVID-19

C. Terracciano, D. Zaino, P. Immovilli, E. Terlizzi, D. Guidetti (Piacenza)

13.20 – 13.25 Creatine kinase and progression rate in amyotrophic lateral sclerosis

M. Ceccanti, C. Cambieri, L. Libonati, E. Onesti, M. Inghilleri (Roma)

13.25 – 13.30 Light-Chain Neurofilaments (Nf-L) assessment in type 3 Spinal Muscular Atrophy patients treated with nusinersen

V. Bozzoni, G. Musso, L. Caumo, L. Bello, S. Tripodi, P. Riguzzi, F. Causin, J. Gabrieli, G. Cester, G. Sorarù, M. Plebani, E. Pegoraro (Padova)

13.30 – 13.35 Improvement of skin biopsy findings after treatment with azatioprina in a case of small fiber neuropathy

R. Milani, ID. Lopez, A. Quattrini, R. Fazio (Milano)

13.35 – 13.40 Discordant clinical outcome in two cousins with X-linked myotubular myopathy

P. D'Ambrosio, M.G. Esposito, S. Morra, A. Palladino, L. Passamano, R. Petillo, E. Picillo, V. Torre, L. Politano (Napoli)

13.40 – 13.45 Validation of the Italian version of the Charcot-Marie-Tooth Health Index (CMT-HI)

C. Pisciotta, E. Ciafaloni, R. Zuccarino, D. Calabrese, P. Saveri, S. Fenu, I. Tramacere, F. Genovese, N. Dilek, N.E. Johnson, C. Heatwole, D.N. Herrmann, D. Pareyson, on behalf of the ACT-CMT study (Milano; Rochester, NY, USA; Iowa City, IA, USA; Arenzano (GE); Bologna; Richmond, VA, USA; Rochester, NY, USA)

13.45 – 13.50 Neuropsychological profile of Becker Muscular Dystrophy

R. Brusa, F. Magri, T. Difonzo, D. Velardo, S. Corti, M. Moggio, M.C. Saetti, G.P. Comi (Milano)

13.50 – 13.55 Long-term respiratory function in SMA type 2 and non-ambulant SMA type 3, longitudinal data from the international SMA consortium (iSMAC)

F. Trucco, M. Scoto, D. Ridout, D. C. De Vivo, B. Darras, E. Bertini, G. Coratti, M. Main, A. Mayhew, J. Montes, R. S. Finkel, E. Mercuri, F. Muntoni on behalf of the international SMA consortium (iSMAC) (Genova; London, UK; New York, USA; Boston, MA, USA; Roma; Newcastle, UK; Orlando, Florida, USA)

13.15 – 13.50 CHANNEL 9 - Emerging neuromuscular entities

13.15 – 13.20 Epidemiological study of HEV prevalence in patients with CIDP and ALS

C. Cambieri, M. Ceccanti, L. Libonati, I. Fiorini, V. Frasca, E. Onesti, G. Taliani, M. Inghilleri (Roma)

13.20 – 13.25 Benign monomelic amyotrophy of upper limb (Hirayama disease): a single center analysis

M.G. Rispoli, L. Ferri, M. Di Pietro, V. Di Stefano, A. Di Muzio (Chieti; Palermo)

13.25 – 13.30 Neurolymphomatosis as the main presentation of relapse of extranodal diffuse large B-cell lymphoma (DLBCL)

M. Campagnolo, M. Cacciavillani, T. Cavallaro, S. Ferrari, L. Pavan, G. Barilà, A. Salvalaggio, F. Castellani, R. Zambello, C. Briani (Padova; Verona)

13.30 – 13.35 An overlapping case of myasthenia gravis, Guillan- Barré syndrome and autoimmune polyglandular syndrome type III

L. Ferri, M.G. Rispoli, V. Falzano, P. Ajdinaj, V. Di Stefano, A. Di Muzio (Chieti; Palermo)

14.00 – 14.15 | Pausa

14.15 - 15.15 | New treatment horizons in the management of SMA

Moderatore – G. Vita

- Recent scientific evidences in the context of early onset SMA | *R. Masson*
- What's new in the landscape of later onset SMA? | *M.C. Pera*

15.15 – 17.15 - Oral communications 3: Biomarkers and outcome measures in neuromuscular disorders

15.15 – 15.30 Long-term functional changes in Becker muscular dystrophy

L. Bello, S. Mastellaro, L. Caumo, P. Riguzzi, V. Zangaro, M. Villa, D. Sabbatini, A. Fusto, B. Merlo, S. Vianello, E. Pegoraro (Padova)

15.30 – 15.45 Sphingomyelin: a novel diagnostic and disease activity biomarker for the management of acquired demyelinating neuropathies

G. Capodivento, C. De Michelis, M. Carpo, R. Fancellu, E. Schirinzi, D. Severi, D. Visigalli, D. Franciotta, G. Siciliano, F. Manganelli, A. Beronio, E. Capello, P. Lanteri, E. Nobile-Orazio, A. Schenone, L. Benedetti, L. Nobbio (Genova; Bergamo; Pisa; Napoli; Pavia; La Spezia; Milano; Rozzano (MI))

15.45 – 16.00 Morphological analysis of TNPO3 and SRSF1 interaction during myogenesis: a super resolution study

R. Costa, M.T. Rodia, N. Zini, V. Pegoraro, R. Marozzo, C. Capanni, C. Angelini, G. Lattanzi, S. Santi, G. Cenacchi (Bologna; Venezia)

16.00 – 16.15 Intraepidermal nerve fiber density as a biomarker of disease severity in hereditary transthyretin amyloidosis with polyneuropathy: data from an Italian cohort

L. Leonardi, A. Truini, A. Fasolino, E. Galosi, M. Luigetti, L. Fionda, F. Vanoli, S. Morino, M. Garibaldi, G. Antonini (Roma)

16.15 – 16.30 Proposal of new functional motor scales to evaluate muscle fatigue in adult SMA patients

G. Ricci, A. Govoni, I. Bortone, L. Billeci, A. Borelli, L. Manca, R. Liguori, M. Coccia, G. Comi, G. Siciliano (Pisa; Milano; Bari; Bologna; Ancona)

16.30 – 16.45 Validation of a new Hand Function Outcome Measure in individuals with Charcot-Marie-Tooth

V. Prada, M. Hamedani, G. Robbiano, G. Mennella, A. Geroldi, A. Zuppa, S. Massucco, R. Zuccarino, L. Mori, E. Bellone, P. Mandich, M. Grandis, A. Schenone (Genova; Coralville, IA, USA)

16.45 – 17.00 Proteomic profiling of cerebrospinal fluid of nusinersen-treated SMA1 patients

M. Sframeli, L. Bianchi, G.L. Vita, R. Oteri, F. Polito, L. Vantaggiato, C. Landi, E. Gitto, S. Messina, L. Bini, M. Aguenouz, G. Vita (Messina; Siena)

17.00 – 17.15 Muscle MRI as a novel outcome measure of hereditary transthyretin amyloidosis: a cross-sectional cohort study

E. Vegezzi, A. Cortese, N. Bergsland, R. Mussinelli, M. Paoletti, F. Solazzo, R. Currò, A. Lozza, X. Deligianni, F. Santini, S. Bastianello, G. Merlini, G. Palladini, L. Obici, A. Pichiecchio (Pavia; London, UK; New York, USA; Pavia; Basel, Switzerland)

17.15 – 18.00 - Oral communications 4: “Innovative therapeutic approaches”

17.15 – 17.30 RNAi therapeutic Patisiran in hATTR amyloidosis: tolerability and management from two centre experience

L. Gentile, M. Russo, M. Luigetti, G. Bisogni, A. Di Paolantonio, A. Romano, V. Guglielmino, M. Sabatelli, A. Toscano, G. Vita, A. Mazzeo (Messina; Roma)

17.30 – 17.45 Pharmacological chaperone to treat myotonia congenita caused by trafficking-defective CIC-1 chloride channel mutants: proof-of-concept with niflumic acid
C. Altamura, E. Conte, D. Sahbani, G.M. Camerino, F. Girolamo, MR. Carratù, P. Imbrici, JF. Desaphy (Bari)

17.45 – 18.00 Inotersen to treat hATTR polyneuropathy: tolerability and management from two centre experience.

M. Luigetti, A. Romano, A. Mazzeo, A. Di Paolantonio, G. Bisogni, V. Guglielmino, M. Russo, L. Gentile, A. Toscano, G. Vita, M. Sabatelli (Roma; Messina)

18.00 - 19.20 | Workshop: Autophagy and other pathogenic mechanisms in NMDs

18.00 - 18.20 IGF-1 mediated signalling to counteract muscle atrophy | *A. Musarò*

18.20 – 18.40 New insights of autophagy regulation and involvement in muscle to neurons communication | *M. Sandri*

18.40 – 19.00 Autophagy in peripheral neuropathies: mechanisms and treatment options
V. Timmerman

19.00 – 19.20 When glycogen becomes insoluble: clinical features and pathobiology of polyglucosan storage disorders | *A. Oldfors*

19.20 - 20.20 | Assemblée AIM e ASNP

Venerdì 11 Dicembre
Streaming on CHANNEL 1

8.30 – 10.00 | Neuromuscular club

8.30 – 8.45 Myoglobinuria as unexpected onset of disease in an asymptomatic 75-years-old man

C. Bonanno, A. Pugliese, G. Nicocia, C. Rodolico, A. Toscano (Messina)

8.45 – 9.00 Brown-Vialetto-Van Laere Syndrome: a clinical report

S. Cotti Piccinelli, F. Novara, B. Risi, E. Baldelli, N. Necchini, A. Galvagni, R. Ciccone, A. Padovani, M. Filosto (Brescia; Pavia)

9.00 – 9.15 Pure neuritic leprosy: a clinical and neuropathological report

N. Necchini, S. Cotti Piccinelli, T. Cavallaro, S. Ferrari, B. Risi, E. Baldelli, A. Padovani, M. Filosto (Brescia; Verona)

9.15 – 9.30 Neuromuscular features in Chorea-Acanthocytosis: a clinical and histopathological report

B. Risi, S. Cotti Piccinelli, F. Novara, E. Baldelli, N. Necchini, A. Galvagni, R. Ciccone, A. Padovani, M. Filosto (Brescia; Pavia)

9.30 – 9.45 A case of intraneurial perineurioma. Limits and advantages of imaging techniques.

M. Romano, D. Coraci, S. Cottone, R. Gasparotti, S. Ferraresi, S. Realmuto, E. Cammarata, L. Padua (Palermo; Roma; Brescia; Rovigo, Milano)

9.45 – 10.00 Adult-onset Krabbe's disease: clinical presentation and characterization of peripheral nerve involvement

M. Tagliapietra, F. Crescenzo, D. Polo, S. Ferrari, T. Cavallaro, G. Zanette, G.M. Fabrizi (Verona; Pescheria del Garda)

10.00 - 11.40 | Workshop:

Innovative therapeutic strategies in genetic neuromuscular diseases (Part II)

10.00 – 10.20 Innovative strategies therapeutic in Muscle dystrophies | *G. P. Comi*

10.20 – 10.40 Advances in the treatment of hereditary ATTR amyloidosis | *A. Mazzeo*

10.40 – 11.00 Challenges in treating CMT: do we see daylight? | *D. Pareyson*

11.00 – 11.20 New Therapeutic approaches in SMA | *E. Mercuri*

11.20 - 11.40 New therapeutic strategies in metabolic myopathies | *O. Musumeci*

11.40 - 12.00 | Pausa

12.00 – 12.45 – POSTER SESSION (Streaming on Channel 2, 3, 4, 5 ,6 ,7 ,8, 9)

12.00 – 12.45 CHANNEL 2 - Innovative therapeutic approaches

12.00 – 12.05 Long-Term Safety and Efficacy of Patisiran: Global Open-label Extension 24-month Data in Patients with Hereditary Transthyretin-mediated Amyloidosis

D. Adams, A. González-Duarte, E. Mauricio, T. Brannagan, T. Coelho, J. Wixner, H. Schmidt, A. Mazzeo, E. Berber, M.T. Sweetser, M.T. White, J.J. Wang, M. Polydefkis (Le Kremlin-Bicêtre, France; Mexico City, Mexico; Jacksonville, FL, USA; New York City, NY, USA; Porto, Portugal; Umeå, Sweden; Muenster, Germany; Messina; Cambridge, USA; Baltimore, USA)

12.05 – 12.10 FIREFISH Part 2: Efficacy and safety of risdiplam (RG7916) in infants with Type 1 spinal muscular atrophy (SMA)

G. Baranello, L. Servais, R. Masson, M. Mazurkiewicz-Będzzińska, K. Rose, D. Vlodayets, H. Xiong, E. Zanolati, M. El-Khairi, S. Fuerst-Recktenwald, M. Gerber, K. Gorni, H. Kletzl, R. Scalco, B. T. Darras on behalf of the FIREFISH Working Group (Milano; London, UK; Liège, Belgium; Oxford, UK; Gdańsk, Poland; Sydney, Australia; Moscow, Russia; Beijing, China; São Paulo, Brazil; Welwyn Garden City, UK; Basel, Switzerland; Boston, MA, USA)

12.10 – 12.15 Systemic Gene Transfer With rAAVrh74.MHCK7.SGCB Increased β -Sarcoglycan Expression in Patients With Limb Girdle Muscular Dystrophy Type 2E

L.R. Rodino-Klapac, E.R. Pozsgai, S. Lewis, D.A. Griffin, A.S. Meadows, P. Roncon, K.J. Lehman, K. Church, N.F. Miller, M.A. Iammarino, L.P. Lowes, J.R. Mendell (Columbus, Ohio, USA; Cambridge, MA, USA)

12.15 – 12.20 Intravenous Onasemnogene Apeparovvec Clinical Development Programs in Spinal Muscular Atrophy (SMA): Integrated Safety Report

D. Chand, R.S. Finkel, E. Mercuri, R. Masson, J. Parsons, R. Cerbini, F. Balducci, A. Kleyn, M. Menier, K. Montgomery, D.M. Sproule, S.P. Reyna, D.E. Feltner, S. Tauscher-Wisniewski, J.R. Mendell (Bannockburn, IL, United States; Orlando, FL, United States; Roma; Milano; Aurora, CO, United States; Columbus, OH, United States)

12.20 – 12.25 Unravelling combined RNA interference and gene therapy in vitro and in vivo disease models as a potential therapeutic strategy for CMT2A

R. De Gioia, M. Nizzardo, S. Bono, S. Salani, V. Melzi, S. Pagliarani, E. Abati, N. Bresolin, G. Comi, S. Corti, F. Rizzo (Milano)

12.25 – 12.30 Testing a novel dasatinib formulation in mdx mouse model: towards the repurposing of Src tyrosine kinase inhibitors in Duchenne muscular dystrophy

F. Sanarica, P. Mantuano, B. Boccanegra, O. Cappellari, E. Conte, G.M. Camerino, A. Cutrignelli, N. Denora, A. Mele, M. De Bellis, A. De Luca (Bari)

12.30 – 12.35 Long-Term Follow-Up (LTFU) of Onasemnogene Apeparvovec Gene Therapy in Spinal Muscular Atrophy Type 1 (SMA1) From Phase 1 START Trial

J.R. Mendell, R. Shell, K.J. Lehman, M. McColly, L.P. Lowes, L.N. Alfano, N.F. Miller, M.A. Iammarino, K. Church, R. Cerbini, S.P. Reyna, F.G. Ogrinc, H. Ouyang, D.M. Sproule, M. Meriggioli, D.E. Feltner, S. Al-Zaidy (Columbus, OH, United States; Bannockburn, IL, United States)

12.35– 12.40 Histopathological features in ambulant patients with Becker Muscular Dystrophy: preliminary data from the Givinstat trial cohort

D. Velardo, M. Ripolone, F. Magri, R. Brusa, A. Govoni, S. Cazzaniga, L. Peverelli, P. Ciscato, S. Zanotti, M. Sciacco, M. Moggio, P. Bettica, G.P. Comi (Milano; Pisa; Lodi)

12.40 – 12.40 SUNFISH Part 2: Efficacy and safety of risdiplam (RG7916) in patients with Type 2 or non-ambulant Type 3 spinal muscular atrophy (SMA)

E. Mercuri, N. Barisic, O. Boespflug-Tanguy, N. Deconinck, A. Kostera-Pruszczyk, R. Masson, E. Mazzone, A. Nascimento, K. Saito, D. Vlodayets, C. Vuillerot, S. Fuerst-Recktenwald, S. Fuhrer, M. Gerber, K. Gorni, H. Kletzl, C. Martin, W.Y. Yeung, J.W. Day on behalf of the SUNFISH Working Group (Roma; Zagreb, Croatia; Paris, France; Gent, Ghent; Brussels, Belgium; Warsaw, Poland; Milano; Barcelona, Spain; Tokyo, Japan; Moscow, Russia; Lyon, France; Basel, Switzerland; Basel, Switzerland; Welwyn Garden City, UK; Palo Alto, CA, USA)

12.00 – 12.40 CHANNEL 3 - Innovative therapeutic approaches

12.00 – 12.05 Ocrelizumab in a case of refractory Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) with antibodies anti Rituximab

S. Casertano, E. Signoriello, F. Rossi, A. Di Pietro, F Tuccillo, S. Bonavita, G. Lus (Napoli)

12.05 – 12.10 Identification and characterization of urine-derived stem cells (USCs) by the novel technology Celector®

M.S. Falzarano, N. Spedicato, A. Margutti, R. El Dani, R. Rossi, S. Zia, P. Reschiglian, B. Roda, A. Grilli, S. Bicciato, A. Ferlini (Ferrara; Bologna; Modena)

12.10 – 12.15 Mutational profile in patients with anti-myelin-associated glycoprotein (MAG) antibody neuropathy identifies new therapeutic target

F. Castellani, A. Visentin, M. Campagnolo, A. Salvalaggio, C. Candiotta, R. Zambello, F. Piazza, L. Trentin, R. Bertorelle, C. Briani (Padova)

12.15 – 12.20 Efgartigimod in Autoimmune Neuromuscular Diseases

A. Guglietta, P. Ulrichs, S. Schmidt, J. Beauchamp, H. de Haard, W. Parys (Zwinjaarde, Belgium)

12.20 – 12.25 CPPs-conjugated antisense nucleotides: a new therapeutic strategy for Spinal Muscular Atrophy symptomatic patients

E. Pagliari, M. Bersani, M. Rizzuti, A. Bordoni, D. Saccomanno, N. Bresolin, GP. Comi, S. Corti, M. Nizzardo (Milano)

12.25 – 12.30 Open-label Study of Patisiran in Patients with hATTR Amyloidosis Post-Orthotopic Liver Transplant

T. Coelho, J. Gillmore, D. Adams, F. Muñoz-Beamud, A. Mazzeo, J. Wixner, V. Planté-Bordeneuve, L. Lladó, S. Arum, J.J. Wang, X. Li, H. Schmid (Porto, Portugal; London, UK; Le Kremlin Bicêtre, France; Huelva, Spain; Messina; Umeå, Sweden; Créteil, France; Barcelona, Spain; Cambridge, MA, USA; Münster, Germany)

12.30 – 12.35 Onasemnogene Apeparvovec Gene Therapy in Presymptomatic Spinal Muscular Atrophy: SPRINT Study Update

K.A. Strauss, M.A. Farrar, K.J. Swoboda, K. Saito, C.A. Chiriboga, R.S. Finkel, S.T. Iannaccone, J.M. Krueger, J.M. Kwon, H.J. McMillan, L. Servais, J.R. Mendell, J. Parsons, M. Scoto, P.B. Shieh, C. Zaidman, F. Baldinetti, M. Schultz, S.P. Reyna, F.G. Ogrinc, S. Kavanagh, D. Chand, D.E. Feltner, S. Tauscher-Wisniewski, B.E. McGill, D.M. Sproule, F. Muntoni (Strasburg, PA, USA; Sydney, NSW, Australia; MA, USA; Tokyo, Japan; New York, NY, USA; Orlando, FL, USA; Dallas, TX, USA; Grand Rapids, MI, USA; Madison, WI, USA; Ottawa, ON, Canada; Oxford, UK; Columbus, OH, USA; Aurora, CO, USA; London, UK; Los Angeles, CA, USA; St. Louis, MO, USA; Bannockburn, IL, USA)

12.35 – 12.40 Long-term impact of inotersen on neuropathy quality of life (QoL) for hereditary transthyretin amyloidosis with polyneuropathy (hATTR-PN): NEURO-TTR open-label extension at 2 years

L. Obici, A. Lovely, T. Coelho, A. Yarlaz, M. Pollock, K. McCausland, I. Conceição, C. Karam, S. Khella, G.Vita, M. Waddington-Cruz, M.V. Llonch (Pavia; Porto, Portugal; Akcea Therapeutics; Lisboa, Portugal; Portland, Oregon, United States; Philadelphia, PA, Unites States; Messina; Rio de Janeiro, Brasil)

12.00 – 12.30 CHANNEL 4 - Current therapies in neuromuscular disorders

12.00 – 12.05 CMV primary infection and GBS, a treatment open question. Report of two cases and review

S. Dallagiacomma, S. Bocci, S. Bartalini, S. Ferrone, L. Franci, F. Ginanneschi, F. Giannini (Siena)

12.05 – 12.10 Systemic Gene Transfer With rAAVrh74.MHCK7.micro-dystrophin in patients with Duchenne Muscular Dystrophy

J.R. Mendell, Z. Sahenk, K. Lehman, C. Nease, L.P. Lowes, N.F. Miller, M.A. Iammarino, L.N. Alfano, J. Vaiea, S. Al-Zaidy, S. Lewis, K. Church, R. Shell, L. Picaro, R.A. Potter, D.A. Griffin, E.R. Pozsgai, M. Hogan, L.R. Rodino-Klapac (Columbus, Ohio, USA; Cambridge, MA, USA)

12.10 – 12.15 Home monitoring of different outcome measures during dose adjustment of intravenous immunoglobulin (IVIg) in patients with chronic inflammatory neuropathies

P.E. Doneddu, D. Mandia, F. Gentile, F. Gallia, G. Liberatore, F. Terenghi, M. Ruiz, E. Nobile-Orazio (Milano)

12.15 – 12.20 Efficacy of Rituximab in refractory Myasthenia gravis

F. Tuccillo, B.M. De Martino, M. Pezzella, M. Esposito, F. Habetswallner (Napoli)

12.20 – 12.25 First and second line treatment in chronic inflammatory demyelinating polyradiculoneuropathy in children: 10 years experience of tertiary pediatric neurology centre

F. Ursitti, L. Papetti, M.A.N. Ferilli, R. Moavero, G. Sforza, F. Vigeveno, M. Valeriani (Roma; Aalborg, Denmark)

12.25 – 12.30 Sub cutaneous Immunoglobulin in two patients affected by Stiff person syndrome: a comparison study with intravenous Immunoglobulin

V. Donadio, E. Fileccia, R. Rinaldi, G.M. Minicuci, R. D'Angelo, L. Bartolomei, R. Liguori (Bologna; Vicenza; Mirano (Ve); Bologna)

12.00 – 12.30 CHANNEL 5 - Disimmune and inflammatory neuromuscular disorders

12.00 – 12.05 Higher muscle damage in patients with anti-Mi2-positive dermatomyositis: a single centre retrospective cohort

F. Girolamo, M. Fornaro, M. Giannini, A. Amati, A. Lia, M. Tampoia, D. Dabbicco, L. Coladonato, M. Trojano, L. Serlenga, F. Iannone (Bari; Strasbourg, France)

12.05 – 12.10 Atypical Acute Motor-Sensitive Axonal Neuropathy (AMSAN) during Acute Hepatitis B Virus (HBV) infection

N. De Angelis, P. Galluzzi, F. Parodi, S. Bocci, F. Ginanneschi, C. Battisti, F. Giannini (Siena)

12.10 – 12.15 Clinico-pathological features in patients with anti-HMGCR immune-mediated necrotizing myopathy: a single-center experience

M. Meneri, D. Velardo, M. Magri, L. Andreoli, A. De Rosa, R. Brusa, M. Mauri, C. Matinato, L. Peverelli, P. Ciscato, S. Corti, C. Cinnante, M. Moggio, M. Sciacco, G.P. Comi (Milano; Lodi)

12.15 – 12.20 Long-term Prognosis in Guillain-Barré Syndrome and Clinical Variants: Focus on Motor and Sensory Outcome

S.G. Grisanti, C. Demichelis, A. Zuppa, V. Prada, A. Beronio, V. Dorindo, A. Schenone, L. Benedetti (Genova; Alessandria; La Spezia)

12.20 – 12.25 Characteristics of corneal innervation in patients with myasthenia gravis

G. Nicocia, C. Bonanno, A. Pugliese, D. Montanini, E. Postorino, S. Messina, G. Vita, P. Aragona, A. Toscano, C. Rodolico (Messina)

12.25 – 12.30 A case of COVID19 associated pharyngeal-cervical-brachial variant of Guillain-Barré Syndrome

G. Liberatore, T. De Santis, P.E. Doneddu, F. Gentile, A. Albanese, E. Nobile-Orazio (Milano)

12.00 – 12.35 CHANNEL 6 - Disimmune and inflammatory neuromuscular disorders

12.00 – 12.05 Anti-AChR Myasthenia Gravis presenting with early predominant left triceps weakness, associated with reversible muscular atrophy

A. Rasera, M. Barillari, D. Cavalli, S. Romito (Verona)

12.05 – 12.10 Isolated cranial neuropathy associated with anti-glycolipid antibodies

C. Petrelli, F. Logullo (Macerata)

12.10 – 12.15 Perivascular M1 macrophages expressing VEGF and SDF1 promote angiogenesis in anti-HMGR immune mediate necrotizing myopathy

F. Girolamo, M. Fornaro, M. Giannini, L. Coladonato, A. Amati, A. Lia, M. Tampoia, D. Dabbicco, R. Tamma, T. Annese, M. Errede, D. Virgintino, D. Ribatti, M. Trojano, F. Iannone, L. Serlenga (Bari; Strasbourg, France)

12.15 – 12.20 Acquired asymmetric brachial plexopathy: a description of a case-series

E. Pezzotti, L. Pasca, A. Gardani, V. Vacchini, E. Rognone, M. Paoletti, G. Cosentino, M. Plumari, C. Cereda, A. Berardinelli (Pavia)

12.20 – 12.25 Myasthenia Gravis and Latent Tuberculosis Infection- A Case Report

M. Sardaro, I. Plasmati, S. Aniello, M. Superbo, D. Liuzzi, S. Altomare, R. Leone R., D. Giorelli D., V. Lucivero (Barletta)

12.25 – 12.30 Report of an unusual course of CANDAs (chronic ataxic neuropathy with disialosyl antibodies)

E. Schirinzi, E. Merico, C. Simoncini, A. Govoni, A. Bacci, R. Calabrese, G. Siciliano (Pisa)

12.30 – 12.35 Inflammation in Children with Neuromuscular Disorders and Sleep Disordered Breathing

F. Trucco, E. Carruthers, J.C. Davies, A. Simonds, A. Bush, H. Tan (Genova; London, UK)

12.00 – 12.30 CHANNEL 7 - Disimmune and inflammatory neuromuscular disorders

12.00 – 12.05 Early neurophysiological abnormalities in Guillain-Barré Syndrome: 4-year experience of Verona center

A. Rasera, S. Romito, A. Segatti, E. Concon, L. Alessandrini, F. Basaldella, A. Badari, C. Arcaro, B. Bonetti, G. Squintani (Verona)

12.05 – 12.10 Severe inflammatory myopathy in a pulmonary carcinoma patient treated with Pembrolizumab: an alert for myologists

D. Velardo, L. Peverelli, A. De Rosa, E. Domina, P. Ciscato, G. Sita, M. Sciacco, M. Moggio, G.P. Comi (Milano; Lodi)

12.10 – 12.15 Recurrent Variant of Miller Fisher Syndrome: is a ganglionopathy? - A Case Report

M. Sardaro, I. Plasmati, C. Santoro, R. Calabrese, S. Aniello, S. Altomare, R. Leone, V. Lucivero (Barletta)

12.15 – 12.20 Subcutaneous immunoglobulins in Myasthenia Gravis and anti-HMGCR myositis
A. Zuppa, C. Demichelis, S.G. Grisanti, C. Cabona, A. Schenone, L. Benedetti, M. Grandis (Genova)

12.20 – 12.25 Childhood Chronic Inflammatory Demyelinating Polyradiculoneuropathy: A Case Series
A. Tozzo, A. Ardisson, C. Ciano, FR. Danti, E. Pagliano, I. Moroni (Milano)

12.25 – 12.30 A case of progressive, stepwise, asymmetrical pure motor neuropathy: MMN versus motor CIDP
S. Tronci, U. Del Carro, M. Filippi, R. Fazio (Milano)

12.00 – 12.35 CHANNEL 8 - Toxic and metabolic neuromuscular disorders

12.00 – 12.05 A diagnostic delay of case of neurogastrointestinal encephalopathy (MNGIE)
F. Aruta, G. Capaldo, C. Pelosi, A. Iovino, M. Giaquinto, P. Romano, R. Rinaldi, S. Trimarco, F. Vitale, L. Ruggiero (Napoli; Avellino; Benevento; Bologna)

12.05 – 12.10 Topiramate in Oxaliplatin-Induced Peripheral Neurotoxicity: more than neuroprotection
P. Alberti, A. Malacrida, S. Semperboni, C. Meregalli, E. Ballarini, G. Cavaletti (Monza; Milano)

12.10 – 12.15 Movement disorders in children with a mitochondrial disease: a cross-sectional survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases
C. Ticci, D. Orsucci, A. Ardisson, E. Bertini, C. Bruno, V. Carelli, D. Diodato, A.M. Donati, M. Filosto, C. La Morgia, C. Lamperti, D. Martinelli, C. Minetti, M. Moggio, T. Mongini, V. Montano, I. Moroni, O. Musumeci, E. Pegoraro, G. Primiano, E. Procopio, A. Rubegni, M. Sciacco, S. Servidei, G. Siciliano, C. Simoncini, P. Tonin, A. Toscano, M. Mancuso, R. Battini, F.M. Santorelli (Pisa; Lucca; Milano; Roma; Genova; Bologna; Firenze; Brescia; Torino; Messina; Padova; Verona)

12.15 – 12.20 Mapping the neurotoxic effect of Oxaliplatin on dorsal root ganglia: MALDI mass spectrometry imaging highlights alterations to the tissue proteome
E. Ballarini, P. Alberti, V. Carozzi, V. Rodriguez-Menendez, A. Smith, G. Cavaletti, F. Magni (Monza)

12.20 – 12.25 Early onset recurrent muscle dysfunction associated with novel POGlut1 mutation
A. Cavaliere, S. Gibertini, A. Ruggieri, F. Blasevich, F.R. Danti, L. Maggi, I. Moroni (Milano)

12.25 – 12.30 Different effects of oxaliplatin and cisplatin incubation on the electrophysiological properties of differentiated F-11 cells
L. Monza, V. Pastori, A. Becchetti, G. Cavaletti, M. Lecchi (Monza; Milano)

12.30 – 12.35 Mitochondrial abnormalities with mtDNA single deletion in association with a LMNA gene frameshift variant: a case report

C. Simoncini, V. Montano, G. Ricci, G. Ali, A. Logerfo, F. Baldinotti, M. Caligo, G. Lattanzi, G. Cenacchi, A. Barison, M. Mancuso, G. Siciliano

12.00 – 12.35 CHANNEL 9 - Toxic and metabolic neuromuscular disorders

12.00 – 12.05 Clinical and genetic study in a patient with Neutral Lipid Storage Disease with Myopathy (NLS-D-M)

E. Baldelli, S. Cotti Piccinelli, B. Risi, N. Necchini, A. Galvagni, A. Padovani, M. Filosto (Brescia)

12.05 – 12.10 Unusual case of motor and dysautonomic neuropathy due to Thallium intoxication

A. Petruzzellis, E. Vecchio, L. Gallicchio, V. Recchia, L. Didonna, P. Lovreglio, G. De Palma, F. Tamma (Bari; Brescia)

12.10 – 12.15 Next generation sequencing-based gene panel tests for the management of diagnosis of lipid myopathies

E.M. Pennisi, F. Cortese, F. Gragnani, L. De Giglio, M. Garibaldi, A. D'Amico, E. Bertini, F. Fattori (Roma)

12.15 – 12.20 Assessment of oxaliplatin-induced peripheral neurotoxicity in different mouse models

E. Pozzi, A. Canta, N. Oggioni, M. Bossi, G. Cavaletti, P. Marmioli (Monza)

12.20 – 12.25 Whole-exome sequencing identifies recessive RDH11 mutations in a new glycogen storage myopathy with retinitis pigmentosa

O. Musumeci, A. Torella, M. Savarese, C. Rodolico, A. Ciranni, R. Oteri, F. Del Vecchio Blanco, G. Esposito, V. Nigro, A. Toscano (Messina; Napoli; Pazzuoli (NA); Helsinki, Finland)

12.25 – 12.30 Evaluation of muscle involvement in Acromegaly and Cushing's syndrome

L. Ruggiero, M.C. De Martino, E. Spina, R. Dubbioso, S. Tozza, C. Pivonello, C. Simeoli, R. Ferrigno, N. Di Paola, R. Iodice, L. Santoro, R. Pivonello, F. Manganelli (Napoli)

12.30 – 12.35 Familial mitochondrial myopathy and renal failure due to biallelic MGME1 mutations

M. Sciacco, M. Ripolone, L. Napoli, D. Piga, D. Velardo, P. Ciscato, M. Moggio, G.P. Comi, D. Ronchi (Milano)

12.45– 13.05 | Clinical and therapeutic updates on Pompe Disease | A. Toscano

13.05 – 14.20 - Oral communications 5: "Emerging neuromuscular entities"

13.05 – 13.20 Biallelic mutations in sord cause a common and potentially treatable neuropathy

A. Cortese, Y. Zhu, A. Rebelo, S. Negri, S. Courel, L. Abreu, C.J. Bacon, Y. Bai, D.M. Bis-Brewer, E. Bugiardini, E. Buglo, M.C. Danzi, S. ME Feely, A. A. Fragkouli, N. A Haridy, R. Isasi, A. Khan, M. Laurà, S. Magri, M. Pipis, C. Pisciotta, E. Powell, A. M. Rossor, J. Sowden, S. Tozza, J. Vandrovcova, J. Dallman, E. Grignani, E. Marchioni, S.S. Scherer, B. Tang, Z. Lin, A. Al-Ajmi, R. Schüle, M. Synofzik, T. Maisonobe, T. Stojkovic, M. Auer-Grumbach, M. A.

Abdelhamed, S.A. Hamed, R. Zhang, F. Manganelli, L. Santoro, P. Saveri, F. Taroni, D. Pareyson, H. Houlden, D.N. Herrmann, M.M. Reilly, M.E. Shy, G. Zhai, S. Zuchner (Miami, Florida, USA; London, UK; Pavia; Iowa City, Iowa, USA; Assiut, Egypt; Milano; Rochester, NY, USA; Napoli, Italy; Coral Gables, Florida, USA; Pavia; Philadelphia, Pennsylvania USA; Hunan Province, China; Al-Jahra, Kuwait; Tübingen, Germany; Paris, France; Vienna, Austria)

13.20 – 13.35 Natural history course of DNM2-related congenital centronuclear myopathy: a retrospective multicentre Italian Study.

D. Diodato, M. Catteruccia, L. Maggi, I. Moroni, E. Pegoraro, P. Riguzzi, L. Ruggiero, M. Garibaldi, A. Berardinelli, M. Pane, G. Atrea, F. Santorelli, E. Bertini, A. D'Amico (Roma; Milano; Padova; Napoli; Pavia; Pisa)

13.35 – 13.50 Biallelic RFC1 expansion is a common cause of idiopathic sensory neuropathy and ganglionopathy

R. Currò, A. Salvalaggio, S. Tozza, C. Gemelli, N. Dominik, V. Galassi Deforie, F. Castellani, E. Vegezzi, S. Efthymiou, G. Cosentino, E. Alfonsi, E. Marchioni, S. Colnaghi, EM. Valente, C. Tassorelli, MM. Reilly, H. Houlden, P. Mandich, E. Bellone, M. Grandis, A. Schenone, L. Santoro, F. Manganelli, C. Briani, A. Cortese (Pavia; Padova; Napoli; Genova; London, UK)

13.50 – 14.05 TYMP mutations result in late onset mitochondrial myopathy with altered muscle mtDNA homeostasis

D. Ronchi, L. Caporali, G. Manenti, M. Meneri, S. Mohamed, M. Contin, D. Piga, V. Mancinelli, S. Corti, M. Sciacco, V. Carelli, G.P. Comi (Milano; Bologna)

14.05 – 14.20 Neuromuscular sarcoidosis: single center experience in 209 consecutive patients.

B. Labella, F. Cinetto, R. Scarpa, R. Manara, E. Pegoraro, C. Agostini, C. Briani (Padova; Treviso)

14.20 - 16.00 | Round table: New suggestions for managing patients with Neuromuscular Disorders

Neuromuscular experts, Patients and Associations

16.00 - 17.00 | Workshop: Paraneoplastic diseases of muscle, NM junction and peripheral nerve: diagnostics and therapy

16.00 – 16.20 Paraneoplastic neuropathies | *S. Ferrari*

16.20 – 16.40 Presynaptic paraneoplastic disorders of the neuromuscular junction: an update | *R. Liguori*

16.40 – 17.00 Paraneoplastic disorders of skeletal muscles | *C. Rodolico*

17.00 – 18.15 | Oral communications 6: “Current therapies in neuromuscular disorders “

17.00 – 17.15 Tubular Aggregate Myopathy caused by activating mutation in STIM1: a functional study in myoblasts and myotubes deriving from affected patients toward the identification of new therapeutic targets

E. Conte, G.M. Camerino, A. Pannunzio, M. Coluccia, M. Mora, L. Maggi, O. Cappellari, A. De Luca, P. Imbrici, A. Liantonio (Bari; Milano)

17.15 – 17.30 An Italian Database to assess the diagnosis, pathogenesis and effect of therapy in Multifocal Motor Neuropathy (MMN) and its variants: a prospective collaborative study

P.E. Doneddu, D. Cocito, A. Mazzeo, L. Benedetti, M. Luigetti, Fazio, C. Briani, G. Siciliano, M. Filosto, G. Antonini, G. Cosentino, R. F. Manganelli, M. Inghilleri, M. Carpo, G.A. Marfia, G.M. Minicuci, L. Gentile, E. Peci, A. Schenone, M. Sabatelli, Tronci, M. Campagnolo, Schirinzi E, G. Liberatore, E. Nobile-Orazio for the MMN Study Group (Milano; Torino; Messina; Genova; La Spezia; Roma; Padova; Pisa; Brescia; Napoli; Roma; Bergamo; Treviglio; Vicenza)

17.30 – 17.45 Nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3

L. Maggi, L. Bello, S. Bonanno, A. Govoni, M. Grandis, L. Passamano, F. Trojsi, F. Cerri, V. Bozzoni, L. Caumo, R. Piras, R. Tanel, E. Saccani, M. Meneri, V. Vacchiano, G. Ricci, E. D’Errico, S. Bortolani, R. Zanin, L. Politano, A. Schenone, S. Previtali, A. Berardinelli, M. Turri, L. Verriello, M. Coccia, R. Liguori, M. Filosto, G. Marrosu, G. Siciliano, I.L. Simone, T. Mongini, G.P. Comi, E. Pegoraro “(Milano; Padova; Pisa; Genova; Napoli; Cagliari; Trento; Parma; Bologna; Bari; Torino; Pavia; Bolzano; Udine; Ancona; Brescia)

17.45 – 18.00 Response to treatment and outcome in late versus early onset MG

F. Pasqualin, S.V. Guidoni, M. Ermani, E. Pegoraro, D.M. Bonifati (Treviso; Padova)

18.00 – 18.15 Short- and long-term efficacy of lenalidomide therapy in patients with POEMS syndrome

F. Terenghi, F. Gentile, F. Gallia, P.E. Doneddu, R. Mazza, M. Ruiz, A. Nozza, E. Nobile-Orazio (Milano; Pavia)

Sabato 12 Dicembre
Streaming on CHANNEL 1

8.30 – 9.30 | Breakfast seminar:

“New perspectives for Myotubular Myopathies”

8.30 – 8.50 XLMTM: clinical spectrum, epidemiology, Nat-His MTM1 Study | *A. D’Amico*

8.50 – 9.10 Histopathology of centronuclear myopathies | *C. Bruno*

9.10 – 9.30 Gene therapy | *W. Müller-Felber*

9.30 – 11.30 | Oral communications 7: “Disimmune and inflammatory neuromuscular disorders”

9.30 – 9.45 Immune-mediated necrotizing myopathies: clinical-serological features of a large Italian cohort of patients

S. Bonanno, M. Lucchini, E. Pancheri, P. Rovere Querini, R. De Lorenzo, A. Biglia, C. Gemelli, P. Riguzzi, L. Bello, A. Pugliese, L. Ruggiero, G. Ricci, C. Fiorillo, G. Vattermi, G. Siciliano, C. Rodolico, M. Filosto, M. Garibaldi, E. Pegoraro, S. Previtalli, P. Tonin, G. Antonini, L. Cavagna, M. Mirabella, L. Maggi (Milano; Roma; Verona; Pavia; Genova; Padova; Messina; Napoli; Pisa)

9.45 – 10.00 Serum pattern of metalloproteinases-2 and-9 and tissue inhibitors in patients with chronic inflammatory demyelinating polyneuropathy

G. Cosentino, V. Di Stefano, M. Montana, E. Alfonsi, C. Tassorelli, F. Brighina, B. Fierro, G. Caimi (Pavia; Palermo)

10.00 – 10.15 Muscle manifestations and CK levels in COVID infection: results of a large cohort of patients inside a Pandemic COVID-19 Area

A. De Rosa, E. Pinuccia Verrengia, I. Merlo, F. Rea, G. Siciliano, G. Corrao, A. Prella (Legnano; Pisa; Milano)

10.15 – 10.30 Guillain-Barré Syndrome and COVID-19: an observational multicenter study from Lombardy and Veneto (Italy)

S. Cotti Piccinelli, S. Gazzina, C. Foresti, B. Frigeni, C. Servalli, M. Sessa, G. Cosentino, E. Marchioni, S. Ravaglia, C. Briani, F. Castellani, G. Zara, F. Bianchi, U. Del Carro, R. Fazio, M. Filippi, E. Magni, G. Natalini, F. Palmerini, A. M. Perotti, A. Bellomo, M. Osio, G. Scopelliti, M. Carpo, A. Raser, G. Squintani, P. E. Doneddu, V. Bertasi, M.S. Cotelli, G.M. Fabrizi, S. Ferrari, L. Bertolasi, F. Ranieri, F. Caprioli, E. Grappa, L. Broglio, G. De Maria, U. Leggio, L. Poli, F. Rasulo, N. Latronico, E. Nobile-Orazio, A. Padovani, A. Uncini, M. Filosto (Brescia; Bergamo; Pavia; Padova; Milano; Treviglio; Verona; Esine; Cremona, Chieti-Pescara)

10.30 – 10.45 Muscle MRI findings and correlation with clinical and immunological parameters in a cohort of IMNM patients

L. Fionda, F. Vanoli, L. Leonardi, J. Alonso Perez, J. Diaz Manera, G. Merlonghi, S. Morino, E. Bucci, G. Alfieri, A. Lauletta, G. Antonini, M. Garibaldi (Roma; Barcelona, Spain)

10.45 – 11.00 Prevalence and relevance of diabetes mellitus in chronic inflammatory demyelinating polyneuropathy

P.E. Doneddu, D. Cocito, F. Manganelli, R. Fazio, C. Briani, M. Filosto, L. Benedetti, E. Bianchi, S. Jann, A. Mazzeo, G. Antonini, G. Cosentino, G.A. Marfia, A. Cortese, A.M. Clerici, M. Carpo, A. Schenone, G. Siciliano, M. Luigetti, G. Lauria, T. Rosso, G. Cavaletti, E. Beghi, G. Liberatore, L. Santoro, E. Spina, E. Peci, S. Tronci, M. Ruiz, S. Cotti Piccinelli, E.P. Verrengia, L. Gentile, L. Leonardi, G. Mataluni, L. Piccolo, E. Nobile-Orazio, on the behalf of the Italian CIDP Database Study Group (Milano; Torino; Napoli; Padova; Brescia; Genova; Messina; Roma; Pavia; Varese; Treviglio; Pisa; Treviso; Monza)

11.00 – 11.15 Distinguishing features of Acute- and Chronic-Onset Chronic Inflammatory Demyelinating Polyradiculoneuropathy

G. Liberatore, F. Manganelli, D. Cocito, R. Fazio, C. Briani, M. Filosto, L. Benedetti, G. Antonini, G. Cosentino, S. Jann, A. Mazzeo, A. Cortese, G. A. Marfia, A. M. Clerici, G. Siciliano, M. Carpo, M. Luigetti, G. Lauria, T. Rosso, G. Cavaletti, P. E. Doneddu, L. Santoro, E. Peci, S. Tronci, M. Ruiz, S. Cotti Piccinelli, A. Schenone, L. Leonardi, A. Toscano, L. Piccolo, G. Mataluni, E. Nobile-Orazio (Milano; Napoli; Torino; Padova; Brescia; Genova; La Spezia; Roma; Palermo; Messina; Pavia; London, UK; Varese; Pisa; Treviglio; Treviso; Monza)

11.15 – 11.30 The neurophysiological lesson from the Italian CIDP database

E. Spina, S. Tozza, P. E. Doneddu, D. Cocito, R. Fazio, C. Briani, M. Filosto, L. Benedetti, G. Cavaletti, S. Jann, A. Mazzeo, G. Antonini, G. Cosentino, G. A. Marfia, A. Cortese, A.M. Clerici, M. Carpo, A. Schenone, G. Siciliano, M. Luigetti, G. Lauria, T. Rosso, G. Liberatore, E. Peci, S. Tronci, M. Ruiz, S. Cotti Piccinelli, E. Pinuccia Verrengia, L. Gentile, L. Leonardi, G. Mataluni, F. Manganelli, L. Santoro, E. Nobile-Orazio (Napoli; Torino; Milano; Padova; Brescia; Genova; Monza; Messina; Roma; Pavia; Varese; Treviglio; Pisa; Treviso)

11.30 - 11.45 | Pausa

11.45- 12.45 | Workshop:

New therapeutic frontiers in dysimmune diseases of muscle, NM junction and peripheral nerve disorders

11.45 – 12.05 Drug-induced worsening in myasthenia gravis” -| *A. Evoli*

12.05 – 12.25 New immunomodulatory treatment for immune-mediated neuropathies | *L. Benedetti*

12.25 – 12.45 State of the art on the therapy of autoimmune myopathies | *M. Mirabella*

12.45 -14.30 | Oral communications 8: “Toxic and metabolic neuromuscular disorders”

12.45 -13.00 Long-term follow up in presymptomatic LOPD patients (PRELOPD STUDY).
An Italian Neuromuscular Centers Experience

O. Musumeci, S. Servidei, T. Mongini, S. Ravaglia, G.P. Comi, F. Santorelli, V. Tugnoli, G. Antonini, E. Pennisi, L. Ruggero, G. Siciliano, C. Sancricca, F. Ricci, R. Brusa, A. Rubegni, E. Sette, M. Garibaldi, G. Ricci, A. Toscano (Messina; Roma; Torino; Pavia; Milano; Pisa; Ferrara; Napoli)

13.00 -13.15 Peripheral Nervous System (PNS) toxicity induced by immune-checkpoint inhibitors in cancer patients: single centre experience

S. Bocci, L. Insana, R. Danielli, F. Ginanneschi, L. Franci, L. Calabrò, A. M. Di Giacomo, M. Maio, F. Giannini (Siena)

13.15 -13.30 Clinical, pathological and prognostic heterogeneity in immune-checkpoint inhibitors-induced myositis

S. Bocci, N. Volpi, R. Danielli, S. Bartalini, L. Calabrò, A.M. Di Giacomo, M. Maio, F. Giannini (Siena)

13.30 -13.45 Neurofilament light chain: a possible serum biomarker for axonal damage in chemotherapy-induced peripheral neurotoxicity rat models

G. Fumagalli, A. Chiorazzi, V. Rodriguez-Menendez, K. Blennow, H. Zetterberg, G. Cavaletti, P. Marmioli (Monza; Mölndal, Sweden; London, UK)

13.45 -14.00 Clinical, laboratory and therapeutic follow-up of large cohort of Late Onset Pompe Disease (LOPD) patients: a single Centre experience

A. Pugliese, G. Nicocia, G. Tavilla, A. Ciranni, R. Oteri, C. Rodolico, G. Vita, O. Musumeci, A. Toscano (Messina)

14.00 -14.15 Arsenic Trioxide-induced peripheral neuropathy: prospective evaluation in a cohort of patients with acute promyelocytic leukemia.

M. Campagnolo, F. Lessi, M. Cacciavillani, M. Riva, A. Salvalaggio, F. Castellani, C. Briani (Padova)

14.15 -14.30 Lipid composition of cellular membranes in Neutral lipid Storage Disease type M: a possible role in etiology of disease?

E.M. Pennisi, D. Tavian, N.I. Noguera, L. DeGiglio, L., M. Mora, L. Maggi, M. Filosto, F. Cortese, M. Garibaldi, S. Missaglia, C. Angelini, E. Palma, A. Macone (Roma; Milano; Brescia; Venezia)

14.30 - 14.50 | Awards and Conclusions