



Congresso Congiunto **AIM - ASNP**

Digital Edition
9 - 12 Dicembre 2020

XX Congresso Nazionale AIM

Associazione Italiana di Miologia

X Congresso Annuale ASNP

Associazione Italiana per lo studio
del Sistema Nervoso Periferico

Con il patrocinio di:

Presidenti del Congresso

Prof. Gian Maria Fabrizi - ASNP

Prof. Carlo Minetti - AIM

Presidente del Comitato Scientifico

Prof. Antonio Toscano

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Paola Mandich

Fiore Manganelli

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Angelo Schenone

Gabriele Siciliano



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PROGRAMME AT GLANCE

Mercoledì 9 Dicembre

14.00 - 14.20	Greetings and introduction
14.20 - 15.20	Workshop: Innovative therapeutic strategies in genetic neuromuscular diseases (Part I)
15.20 - 16.40	Workshop: New tools in the management of patients with neuromuscular disorders
16.40 - 16.50	Break
16.50 - 18.20	Oral communications 1: "Updating diagnostic tools in neuromuscular disorders"
18.20 - 18.50	Lecture: Neuromuscular disorders prevention: newborn screening, prenatal and pre-implantation diagnosis and counseling
18.50 - 20.10	Workshop: Innovative diagnostic aspects in neuromuscular disorders

Giovedì 10 Dicembre

08.30 - 09.30	Breakfast Seminar: Transthyretin-related Amyloidosis
09.30 - 11.30	Oral communications 2: "Inherited neuromuscular disorders"
11.30 - 12.00	Lecture: "High-cost drugs economic sustainability for the National Health Service. How long can we afford them?"
12.00 - 12.15	Break

PROGRAMME AT GLANCE

Giovedì 10 Dicembre

12.15 - 13.15	Workshop: Imaging techniques in muscle and peripheral nerve diseases	
13.15 - 14.00	Poster session	
CH. 2 Updating diagnostic tools in neuromuscular disorders	CH. 3 Inherited neuromuscular disorders	CH. 4 Inherited neuromuscular disorders
CH. 5 Inherited neuromuscular disorders	CH. 6 Biomarkers and outcome measures in neuromuscular disorders	CH. 7 Biomarkers and outcome measures in neuromuscular disorders
CH. 8 Biomarkers and outcome measures in neuromuscular disorders	CH. 9 Emerging neuromuscular entities	
14.00 - 14.15	Break	
14.15 - 15.15	New treatment horizons in the management of SMA	
15.15 - 16.15	Oral communications 3: "Biomarkers and outcome measures in neuromuscular disorders"	
16.15 - 17.15	Oral communications 4: Biomarkers and outcome measures in neuromuscular disorders	
17.15 - 18.00	Oral communications 5: "Innovative therapeutic approaches"	
18.00 - 19.20	Workshop: Autophagy and other pathogenic mechanisms in NMDs	

Venerdì 11 Dicembre

08.30 - 10.00	Neuromuscular club
10.00 - 11.40	Workshop: Innovative therapeutic strategies in genetic neuromuscular diseases (Part II)
11.40 - 12.00	Break

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PROGRAMME AT GLANCE

Venerdì 11 Dicembre

12.00 - 12.45	Poster session		
CH. 2 Innovative therapeutic approaches	CH. 3 - Innovative therapeutic approaches	CH. 4 Current therapies in neuromuscular disorders	
CH. 5 Disimmune and inflammatory neuromuscular disorders	CH. 6 Disimmune and inflammatory neuromuscular disorders	CH. 7 Disimmune and inflammatory neuromuscular disorders	
CH. 8 Toxic and metabolic neuromuscular disorders	CH. 9 Toxic and metabolic neuromuscular disorders		
12.45 - 13.05	Clinical and therapeutic updates on Pompe Disease		
13.05 - 14.20	Oral communications 6: “Emerging neuromuscular entities”		
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		
17.00 - 18.15	Oral communications 7: “Current therapies in neuromuscular disorders “		

Sabato 12 Dicembre

08.30 - 09.30	Breakfast seminar: "New perspectives for Myotubular Myopathies"
09.30 - 11.30	Oral communications 8: "Disimmune and inflammatory neuromuscular disorders"
11.30 - 11.45	Break

Congresso Congiunto

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PROGRAMME AT GLANCE

Sabato 12 Dicembre

11.45 - 12.45

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

12.45 - 14.30

Oral communications 9:
"Toxic and metabolic neuromuscular disorders"

14.30 - 14.50

Awards and Conclusions





Programme

14.00 - 14.20 | **Greetings and introduction**

Prof. Gian Maria Fabrizi - ASNP

Prof. Carlo Minetti - AIM

Prof. Antonio Toscano - Presidente del Comitato Scientifico

14.20 - 15.20 | **Workshop:**

Innovative therapeutic strategies in genetic neuromuscular diseases (Part I)

Chairs: M. Grandis, L. Maggi

14.20 - 14.40 | An "antisense" approach for neuromuscular diseases | *T. 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

Chairs: M. Sabatelli, G. Siciliano

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 | Computational 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 | **Break**

16.50 - 18.20 | **Oral communications I: "Updating diagnostic tools in neuromuscular disorders"**

Chairs: A. Ferlini, M. Savarese

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, V. Sansone, A. Barp,
E. Albamonte, T. Evangelista, A. Ferlini (Ferrara; Messina; Roma; Milano; 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. Bensemmame, A. Sbatillo, L. Bello, E. Pegoraro, A. Ferlini
(Ferrara; Shenzhen, China; Zadar, Croatia; Alger; Kharkiv, Ukraine; Padova)*

Mercoledì 9 Dicembre

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 - Chair: P. Mandich

18.50 - 20.10 | **Workshop:**

Innovative diagnostic aspects in neuromuscular disorders

Chairs: E. Pegoraro, G. Lauria

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

08.30 - 9.30 | **Breakfast Seminar: Transthyretin-related Amyloidosis**

Chairs: G. Antonini, M. Lugetti

08.30 - 8.50 | Molecular mechanisms | L. Obici

08.50 - 9.10 | Clinical presentation, diagnosis and treatment of peripheral neuropathy | G. Vita

09.10 - 9.30 | Clinical and therapeutic aspects of cardiomyopathy | G. Limongelli

09.30 - 11.30 | **Oral communications 2 :**

"Inherited neuromuscular disorders"

Chairs: E. Bellone, G. Ricci

09.30 - 09.45 | Clinical, morphological and genetic data of patients with distal and myofibrillar myopathies: report from the Italian network

S. Bortolani, S. Bonanno, G. Vattemi, 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)

09.45 - 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 CMT2I patients carrying the MPZ P70S mutation](#)
P. Saveri, C. Pisciotto, 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. Vattemi, 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 - Chair: P. Masciandro
- 12.00 - 12.15 | **Break**

12.15 -13.15 | **Workshop:**

Imaging techniques in muscle and peripheral nerve diseases - *Chairs: M. Romano, G. Tasca*

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** (Sessione non accreditato ECM)

• **CHANNEL 2** - Updating diagnostic tools in neuromuscular disorders | *Chairs: A. Cortese, L. Ruggiero*

• **CHANNEL 3** - Inherited neuromuscular disorders | *Chairs: F. Cotti Piccinelli, C. Pisciotta*

• **CHANNEL 4** - Inherited neuromuscular disorders | *Chairs: L. Losito, S. Tozza*

• **CHANNEL 5** - Inherited neuromuscular disorders | *Chairs: G. Lattanzi, S. Tamburin*

• **CHANNEL 6** - Biomarkers and outcome measures in neuromuscular disorders | *Chairs: L. Nobbio, A. Trabacca*

• **CHANNEL 7** - Biomarkers and outcome measures in neuromuscular disorders

Chairs: S. Mariotto, C. Semplicini

• **CHANNEL 8** - Biomarkers and outcome measures in neuromuscular disorders

Chairs: P.E. Doneddu, A. Pugliese

• **CHANNEL 9** - Emerging neuromuscular entities | *Chairs: A. Ardisson, C. Taveggia*

14.00 - 14.15 | **Break**

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

Chairs: E. Bertini, GL 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 - 16.15 | **Oral communications 3:**

"Biomarkers and outcome measures in neuromuscular disorders"

Chairs: G. Piscosquito, L. Politano

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. Luigietti, L. Fionda, F. Vanoli, S. Morino, M. Garibaldi, G. Antonini (Roma)
- 16.15 - 17.15 | **Oral communications 4:**
“Biomarkers and outcome measures in neuromuscular disorders”
Chairs: C. Angelini, A. Uncini
- 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. Pichiechio (Pavia; London, UK; New York, USA; Pavia; Basel, Switzerland)
- 17.15 - 18.00 | **Oral communications 5:**
“Innovative therapeutic approaches”
Chairs: S. Corti, A. Schenone
- 17.15 - 17.30 | [RNAi therapeutic Patisiran in hATTR amyloidosis: tolerability and management from two centre experience](#)
L. Gentile, M. Russo, M. Luigietti, 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. Sabbani, G.M. Camerino, F. Girolamo, MR. Carratù, P. Imbriici, JF. Desaphy (Bari)
- 17.45 - 18.00 | [Inotersen to treat hATTR polyneuropathy: tolerability and management from two centre experience](#)
M. Luigietti, A. Romano, A. Mazzeo, A. Di Paolantonio, G. Bisogni, V. Guglielmino, M. Russo, L. Gentile, A. Toscano, G. Vita, M. Sabatelli (Roma; Messina)

Giovedì 10 Dicembre

18.00 - 19.20 | **Workshop:**

Autophagy and other pathogenic mechanisms in NMDs

Chairs: A. Bolino, M. Moggio

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*

Venerdì 11 Dicembre

08.30 - 10.00 | Neuromuscular club

Chairs: L. Santoro, S. Servidei

08.30 - 08.45 | **Myoglobinuria as unexpected onset of disease in an asymptomatic 75-years-old man**

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

08.45 - 09.00 | **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)

09.00 - 09.15 | **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)

09.15 - 09.30 | **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)

09.30 - 09.45 | **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)

09.45- 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)

Chairs: F. Manganelli, A. Martinuzzi

- 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 | **Break**

12.00 - 12.45 | **Poster session (Sessione non accreditato ECM)**

- **CHANNEL 2** - Innovative therapeutic approaches | *Chairs: J. Desafy, L. Gentile*
- **CHANNEL 3** - Innovative therapeutic approaches | *Chairs: R. Fazio, A. Petrucci*
- **CHANNEL 4** - Current therapies in neuromuscular disorders | *Chairs: F. Ricci, M. Russo*
- **CHANNEL 5** - Disimmune and inflammatory neuromuscular disorders
Chairs: G. Nicocia, V. Provitera
- **CHANNEL 6** - Disimmune and inflammatory neuromuscular disorders | *Chairs: C. Gemelli, A. Pini*
- **CHANNEL 7** - Disimmune and inflammatory neuromuscular disorders | *Chairs: G. Astrea, D. Coraci*
- **CHANNEL 8** - Toxic and metabolic neuromuscular disorders | *Chairs: P. Alberti, S. Ravaglia*
- **CHANNEL 9** - Toxic and metabolic neuromuscular disorders | *Chairs: M. Garnero, M. Sciacco*

12.45 - 13.05 | Clinical and therapeutic updates on Pompe Disease | *A. Toscano - Chair: C. Minetti*

13.05 - 14.20 | **Oral communications 6: "Emerging neuromuscular entities"**

Chairs: M. Mancuso, I. Moroni

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. Bugiardi, 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. Maissonobe, T. Stojkovic, M. Auer-Grumbach, M. A. Abdelbamed, 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-Jabra, 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

Discussants: T. Mongini, D. Pareyson, V. Sansone, A. Schenone

16.00 - 17.00 | **Workshop:**

Paraneoplastic diseases of muscle, NM junction and peripheral nerve: diagnostics and therapy

Chairs: C. Lamperti, A. Quattrini

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 7:

“Current therapies in neuromuscular disorders”

Chairs: A. Berardinelli, D. Cocito

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)

**08.30 - 09.30 | Breakfast seminar:
“New perspectives for Myotubular Myopathies”**

Chairs: A. Di Muzio, R. Massa

08.30 - 08.50 | XLMTM: clinical spectrum, epidemiology, Nat-His MTM1 Study | *A. D'Amico*

08.50 - 09.10 | Histopathology of centronuclear myopathies | *C. Bruno*

09.10 - 09.30 | Gene therapy | *W. Müller-Felber*

**09.30 - 11.30 | Oral communications 8:
“Disimmune and inflammatory neuromuscular disorders”**

Chairs: C. Briani, G. Meola

09.30 - 09.45 | 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. Vattemi, G. Siciliano, C. Rodolico, M. Filosto, M. Garibaldi, E. Pegoraro, S. Previtali, P. Tonin, G. Antonini, L. Cavagna, M. Mirabella, L. Maggi
(Milano; Roma; Verona; Pavia; Genova; Padova; Messina; Napoli; Pisa)*

09.45 - 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. Rasera, 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 | **Break**

11.45 - 12.45 | **Workshop:**

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

Chairs: M. Filosto, E. Nobile-Orazio

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 9: “Toxic and metabolic neuromuscular disorders”

Chairs: G. Cavaletti, M. Zeviani

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. Marmiroli (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. Maccone (Roma; Milano;
Brescia; Venezia)*

14.30 - 14.50 | Awards and Conclusions

9 - 12 Dicembre 2020

Congresso Congiunto

AIM - ASNP

XX Congresso Nazionale AIM

Associazione Italiana di Miologia

X Congresso Annuale ASNP

Associazione Italiana per lo studio
del Sistema Nervoso Periferico



POSTER SESSIONS

POSTER SESSION 1 - Giovedì 10 Dicembre 13.15 - 14.00

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

Chairs: A. Cortese, L. Ruggiero

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. Biani (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. Picchiechio, 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; Bosisto 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 -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)

POSTER SESSION 1

13.15 - 14.00 CHANNEL 3 - Inherited neuromuscular disorders

Chairs: C. Pisciotta, S. Cotti Piccinelli

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 - Tooths 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)

POSTER SESSION 1

13.15 - 14.00 - CHANNEL 4 - Inherited neuromuscular disorders

Chairs: L. Losito, S. Tozza

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, arthrogryposis 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. Vattemi (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)

POSTER SESSION 1

13.15 - 14.00 CHANNEL 5 - Inherited neuromuscular disorders

Chairs: G. Lattanzi, S. Tamburin

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. Luigietti, 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. Baucé, 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; Bethesda, MD, USA; Catania)

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)

POSTER SESSION 1

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

Chairs: L. Nobbio, A. Trabacca

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. Pisciotta, 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

Chairs: S. Mariotto, C. Semplicini

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. Vattemi, 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)

POSTER SESSION 1

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

F. Saccà, S. Berrib-Aknin, K.G. Claey's, R. Mantegazza, H. Murai, E. Bagsbaw, 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. Baldinetti, 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

Chairs: P.E. Doneddu, A. Pugliese

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 azathioprina in a case of small fiber neuropathy

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

POSTER SESSION 1

13.35 - 13.40 Validation of the Italian version of the Charcot-Marie-Tooth Health Index (CMT-HI)

C. Pisciotto, 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)

13.40 - 13.45 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)

13.45 - 13.50 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. Maybew, 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 - 13.35 CHANNEL 9 - Emerging neuromuscular entities

Chairs: A. Ardisson, C. Taveggia

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)

POSTER SESSION 2 - Venerdì 11 Dicembre 12.00- 12.45

12.00 - 12.45 CHANNEL 2 - Innovative therapeutic approaches

Chairs: J. Desafy, L. Gentile

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-Beldzinska, K. Rose, D. Vlodavets, H. Xiong, E. Zanoteli, 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; Gdansk, 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 Apeparvovec 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. Baldinetti, 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)

POSTER SESSION 2

12.35- 12.40 Histopathological features in ambulant patients with Becker Muscular Dystrophy: preliminary data from the Givinostat 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 - 12.45 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. Vlodavets, 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

Chairs: R. Fazio, A. Petrucci

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. Biciato, 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. Candiotti, 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 (Zwijnjaarde, 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, G.P. 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)

POSTER SESSION 2

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. Baladinetti, 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. Yarlus, M. Pollock, K. McCausland, I. Conceição, C. Karam, S. Kbella, G. Vita, M. Waddington-Cruz, M.V. Llonch
(Pavia; Porto, Portugal; Akcea Therapeutics; Lisboa, Portugal; Portland, Oregon, United States; Philadelphia, PA, United States; Messina; Rio de Janeiro, Brasil)

12.00 - 12.30 CHANNEL 4 - Current therapies in neuromuscular disorders

Chairs: F. Ricci, M. Russo

12.00 - 12.05 [CMV primary infection and GBS, a treatment open question. Report of two cases and review](#)

S. Dallagiacoma, 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. Sabenk, 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)

POSTER SESSION 2

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

Chairs: G. Nicocia, V. Provitera

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

Chairs: C. Gemelli, A. Pini

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)

POSTER SESSION 2

12.10 - 12.15 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)

12.15 - 12.20 Early neurophysiological abnormalities in Guillain-Barré Syndrome: 4-year experience of Verona center

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

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. Giorrelli D., V. Lucivero (Barletta)

12.25 - 12.30 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)

12.30 - 12.35 Perivascular M1 macrophages expressing VEGF and SDF1 promote angiogenesis in anti-HMGCR immune mediate necrotizing myopathy

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

12.00 - 12.30 CHANNEL 7 - Disimmune and inflammatory neuromuscular disorders

Chairs: G. Astrea, D. Coraci

12.00 - 12.05 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)

12.05 - 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 Report of an unusual course of CANDA (chronic ataxic neuropathy with disialosyl antibodies)

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

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. Ardissonne, C. Ciano, FR. Danti, E. Pagliano, I. Moroni (Milano)

POSTER SESSION 2

12.25 - 12.30 A case of progressive, stepwise, asymmetrical pure motor neuropathy: MMN versus motor CIPD

S. Tronci, U. Del Carro, M. Filippi, R. Fazio (Milano)

12.00 - 12.35 CHANNEL 8 - Toxic and metabolic neuromuscular disorders

Chairs; P. Alberti, S. Ravaglia

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. Servadei, 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 POGUT1 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



POSTER SESSION 2

12.00 - 12.35 CHANNEL 9 - Toxic and metabolic neuromuscular disorders

Chairs: M. Garnero, M. Sciacco

12.00 - 12.05 [Clinical and genetic study in a patient with Neutral Lipid Storage Disease with Myopathy \(NLSD-M\)](#)

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. Marmiroli (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)

9 - 12 Dicembre 2020

Congresso Congiunto

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Informazioni generali

Lingue ufficiali del Congresso: Italiano e Inglese

I Workshop in cui sono presenti relatori stranieri dovranno essere discussi esclusivamente in lingua inglese.

Le diapositive delle presentazioni, abstract e poster dovranno essere in lingua inglese

Gli strumenti della piattaforma congressuale:

- **Timetable:** in questa sezione sarà possibile trovare orario e canale in cui saranno trasmesse le sessioni del Congresso. Alla fine dell'evento sarà possibile trovare in questa sezione le presentazioni pre-registrate on-demand
- **Agenda Personale:** in questa sezione sarà possibile creare una propria agenda personale, selezionando le sessioni dalla sezione timetable, semplicemente con un click, dopo il quale si attiveranno le notifiche sia email che della piattaforma stessa per essere avvisati dell'inizio imminente della sessione.
- **Lounge area:** area libera in cui è possibile vedere tutti gli utenti collegati e consultare le informazioni base inserite nel profilo. Una chat pubblica consentirà un coinvolgimento globale, mentre sarà possibile inviare una richiesta di contatto che, se accettata, consentirà di aprire una chat privata o una videocall
- **Account:** in questa sezione si potrà completare le proprie informazioni e decidere se si vorrà condividere le proprie informazioni personali con gli altri partecipanti
- **Comitato Scientifico e lista relatori:** in queste due sezioni sarà possibile visualizzare i membri del Comitato Scientifico e i relatori coinvolti
- **Area sponsor:** area dedicata alle aziende sponsor dove sarà possibile fruire dei contenuti messi a disposizione.

Sessioni LIVE:

Le sessioni andranno in onda secondo data e orario come da programma scientifico.

Il pubblico online avrà la possibilità di interagire direttamente con i Relatori tramite uno strumento di chat. I moderatori delle sessioni selezioneranno le domande dal pubblico e le presenteranno al relatore/presentatore alla fine di ogni intervento.

On-demand:

Le presentazioni preregistrate saranno accessibili alla fine del Congresso e disponibili fino al 30 maggio 2021 nella sezione Timetable in corrispondenza di ciascuna sessione

Premiazioni

Sabato 12 Dicembre alle ore 14.30 è prevista una sessione nella quale saranno assegnati dei premi per il miglior poster e comunicazione orale.

Il premio sarà assegnato soltanto al presentatore, under 41 anni, che sarà presente on line al momento della premiazione.

Attestato partecipazione

Sarà possibile ottenere il proprio attestato di partecipazione generandolo autonomamente dalla sezione account nella tab "Attestato".

Atti del Congresso:

Gli atti del Congresso sono disponibili on line sulla rivista Neurological Sciences tramite il seguente link:
<https://rdcu.be/cbxEN>

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Numero di credito ECM assegniati: 6

L'evento è stato accreditato ECM per le seguenti professioni e discipline

Professione	Discipline
Farmacista:	<i>Farmacia Ospedaliera; Farmacia Territoriale;</i>
Biologo:	<i>Biologo;</i>
Logopedista:	<i>Logopedista;</i>
Terapista Occupazionale:	<i>Terapista Occupazionale;</i>
Psicologo:	<i>Psicoterapia; Psicologia;</i>
Medico Chirurgo:	<i>Cardiologia; Genetica Medica; Malattie Metaboliche e Diabetologia; Malattie Dell'apparato Respiratorio; Medicina Fisica e Riabilitazione; Neonatologia; Neurologia; Neuropsichiatria Infantile; Pediatria; Chirurgia Pediatrica; Neurochirurgia; Ortopedia e Traumatologia; Anestesia e Rianimazione; Laboratorio di Genetica Medica; Neurofisiopatologia; Neuroradiologia; Pediatria (pediatri di libera scelta); Scienza dell'alimentazione e dietetica; Medicina Interna</i>
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Fisioterapista:	<i>Fisioterapista</i>
Infermiere:	<i>Infermiere</i>
Dietista:	<i>Dietista;</i>
Tecnico di Neurofisiopatologia	<i>Tecnico di Neurofisiopatologia</i>
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Avranno diritto ai crediti ECM solo coloro che:

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- avranno superato il questionario di apprendimento finale che sarà disponibile al termine del Congresso fino a 72h, entro le ore 15.00 del giorno 16 dicembre (è concesso un solo tentativo)
- avranno compilato la scheda anagrafica disponibile sulla piattaforma ECM del congresso (è concesso un solo tentativo)
- avranno compilato la scheda di valutazione disponibile sulla piattaforma ECM del congresso



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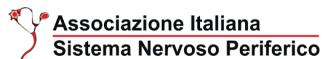
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