

XXI Congresso Nazionale

Associazione Italiana di Miologia

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2021**

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XXI Congresso Nazionale

Associazione Italiana di Miologia



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**CONSIGLIO
DIRETTIVO**
ASSOCIAZIONE
ITALIANA DI MIOLOGIA



XXI Congresso Nazionale

Associazione Italiana di Miologia

Consiglio Direttivo

Associazione Italiana di Miologia

Prof. Carlo Minetti - Presidente

Prof. Gabriele Siciliano - Past President

Prof. Carmelo Rodolico - Segretario

Dr. Luca Bello - Tesoriere

Consiglieri

Prof. Giovanni Antonini

Dr.ssa Adele D'Amico

Prof. Vincenzo Nigro

Dr. Antonio Di Muzio

Prof. Rocco Liguori

Prof. Giacomo Pietro Comi

www.miologia.org

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Associazione Italiana di Miologia



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FACULTY

Emilio Albamonte, *Milano*
Anna Ambrosini, *Milano*
Corrado Angelini, *Padova*
Giovanni Antonini, *Roma*
Alberto Auricchio, *Napoli*
Rita Barresi, *Venezia*
Luca Bello, *Padova*
Angela Berardinelli, *Pavia*
Enrico Bertini, *Roma*
Claudio Bruno, *Genova*
Giacomo Pietro Comi, *Milano*
Stefania Corti, *Milano*
Grazia Crescimanno, *Palermo*
Adele D'Amico, *Roma*
Grazia D'Angelo, *Bosisio Parini (Lecco)*
Annamaria De Luca, *Bari*
Antonio Di Muzio, *Chieti*
Maria Alice Donati, *Firenze*
Victor Dubowitz, *Roma*
Amelia Evoli, *Roma*
Alessandra Ferlini, *Ferrara*
Massimiliano Filosto, *Brescia*
Chiara Fiorillo, *Genova*
Delia Gagliardi, *Milano*
Fabrizio Gardoni, *Milano*
Matteo Garibaldi, *Roma*
Michela Guglieri, *Newcastle (UK)*
Costanza Lamperti, *Milano*
Steven Laurie, *Barcelona (ES)*
Rocco Liguori, *Bologna*
Lorenzo Maggi, *Milano*
Francesca Magri, *Milano*
Michelangelo Mancuso, *Pisa*
Renato Mantegazza, *Milano*
Roberto Massa, *Roma*
Giovanni Meola, *Milano*
Eugenio Mercuri, *Roma*
Carlo Minetti, *Genova*
Federico Mingozzi, *Evry Cedex (FR)*
Massimiliano Mirabella, *Roma*
Maurizio Moggio, *Milano*
Cristina Molera, *Barcelona (ES)*
Tiziana Mongini, *Torino*
Antonio Musarò, *Roma*
Olimpia Musumeci, *Messina*
Vincenzo Nigro, *Napoli*
Ichizo Nishino, *Tokyo (JP)*
Elena Pegoraro, *Padova*
Antonella Pini, *Bologna*
Luisa Politano, *Napoli*
Stefano Previtali, *Milano*
Guido Primiano, *Roma*
Marco Rasconi, *Milano*
Sabrina Ravaglia, *Pavia*
Giulia Ricci, *Pisa*
Enzo Ricci, *Roma*
Carmelo Rodolico, *Messina*
Dario Ronchi, *Roma*
Valeria Sansone, *Milano*
Filippo Maria Santorelli, *Pisa*
Monica Sciacco, *Milano*
Serenella Servidei, *Roma*
Gabriele Siciliano, *Pisa*
Giorgio Tasca, *Roma*
Francesco Danilo Tiziano, *Roma*
Paola Tonin, *Verona*
Antonio Toscano, *Messina*
Antonio Trabacca, *Brindisi*
Rossella Tupler, *Modena*
Daniele Velardo, *Milano*
Emilie Venereau, *Milano*
Nicol Voermans, *Nijmegen (NL)*

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PROGRAMME AT A GLANCE

Wednesday, 1st December 2021

14.00	Conference opening
14.10 - 14.30	Introduction
14.30 - 16.20	Workshop 1: Neuromuscular diseases during and after the COVID-19 Pandemic
16.20 - 17.20	Round Table: New nomenclature for LGMD. Is it exhaustive? New nomenclature for LGMD. Is it exhaustive?
17.20 - 18.50	Workshop 2 – Basic Muscle Research

Thursday, 2nd December 2021

08.00 - 09.00	Early Morning Seminar: Therapies for Myasthenia Gravis and Myasthenic Syndromes
09.00 - 10.40	Workshop 3: New treatment approaches for neuromuscular diseases
10.40 - 12.40	Symposium: Update of Pharmacological Treatment of SMA
12.40 - 13.10	Hot Topics
14.00 - 15.15	ORAL COMMUNICATIONS 1: New treatments for SMA
17.00 - 18.45	17.00-18.45 ORAL COMMUNICATIONS 2 - Myopathies and muscular dystrophies: new phenotypes, biomarkers, and therapies – 1

PROGRAMME AT A GLANCE

15.15 – 17.00	Poster session (Sessione non accreditata ECM)	
	CH. 1 Dystrophinopathies and muscular dystrophies	CH. 2 Myasthenia, inflammatory myopathies, channelopathies
	CH. 3 Myopathies	CH. 4 SMA and motor neuron disorders

17.00 – 18.45	17.00-18.45 ORAL COMMUNICATIONS 2 - Myopathies and muscular dystrophies: new phenotypes, biomarkers, and therapies – 1
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Friday, 3rd December 2021

08.00 – 08.40	Early Symposium: Myotubular Myopathy (MTM)
08.40 – 10.00	Workshop 4 - Update on mitochondrial disease
10.00 – 10.30	Invited Lecture: Latest developments in Facioscapulohumeral Dystrophy
10.30 – 11.30	Workshop 5 - Inflammatory myopathies
11.30 – 12.00	Invited Lecture “Recent update on dermatomyositis”
12.00 – 13.00	Symposium - Patient assessment in view of new treatments for Pompe disease
14.00 – 15.30	ORAL COMMUNICATIONS 3 Myopathies and muscular dystrophies: new phenomic, genomic, and pathogenetic aspects

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PROGRAMME AT GLANCE

Friday, 3rd December 2021

15.30 - 17.15

ORAL COMMUNICATIONS 4

Natural history and new diagnostic tools in neuromuscular disorders

17.15 - 18.45

ORAL COMMUNICATIONS 5

Myopathies and muscular dystrophies: new phenotypes, biomarkers, and therapies – 2

Saturday, 4th December 2021

08.30 - 09.30

Wake up Muscle Club

09.30 - 11.00

Workshop 6 - Next generation molecular diagnostics

11.00 - 12.30

AIM meets Patients and Family Association

12.30

Awards Ceremony for the best works

13.00

Conclusions



ASSOCIAZIONE ITALIANA
MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

**XXI Congresso
Nazionale**

Associazione Italiana di Miologia

PROGRAMME

Wednesday, 1st December 2021

14.00 | Conference opening

Greetings from the Authorities and introduction of the AIM President

14.10 - 14.30 | Introduction: *Carlo Minetti*

Muscle biopsy: is it still a modern approach? Cinderella and to-day's Myology

Guest: *Victor Dubowitz*

14.30 - 16.20 | Workshop 1:

Neuromuscular diseases during and after the COVID-19 Pandemic

Chairmen: *Giacomo Comi - Rocco Liguori*

- Muscular complications | *Delia Gagliardi*
- Peripheral nerve complications | *Massimiliano Filosto*
- Neuromuscular plate junction complications | *Rocco Liguori*
- Telemedicine and patient follow-up | *Giulia Ricci*
- Territorial management of neuromuscular patients | *Antonio Trabacca*

16.20 - 17.20 | Round Table

New nomenclature for LGMD. Is it exhaustive?

Chairman: *Corrado Angelini*

Participants: *Giacomo Comi, Carlo Minetti, Elena Pegoraro, Vincenzo Nigro, Antonio Toscano*

17.20 - 18.50 | Workshop 2

Basic Muscle Research

Chairmen: *Elena Pegoraro, Claudio Bruno*

- Muscle homeostasis and regeneration: from cellular and molecular mechanisms to therapeutic opportunities | *Antonio Musarò*
- SMA molecular pathogenesis downstream of the SMN1 gene | *Stefania Corti*
- The alarmin HMGB1 links oxidative stress and inflammation in muscular dystrophy | *Emilie Venereau*
- Mechanical-metabolic dysfunction in muscle disorders: new insights and therapeutic implications | *Annamaria De Luca*

Thursday, 2nd December 2021

08.00 - 09.00 | Early Morning Seminar:

Therapies for Myasthenia Gravis and Myasthenic Syndromes

Chairman: *Antonio Di Muzio*

- Atypical clinical Phenotypes | *Carmelo Rodolico*
- Focus on complex patients in Myasthenia gravis | *Amelia Evoli*
- New Therapeutical approaches in Myasthenia | *Renato Mantegazza*

09.00 - 10.40 | **Workshop 3**

New treatment approaches for neuromuscular diseases

Chairmen: *Gabriele Siciliano, Maurizio Moggio*

- Molecular therapy in muscular dystrophy | *Stefano Previtali*
- Long-term effects of innovative treatments in DMD/BMD | *Luca Bello*
- Myotonia and channelopathies | *Lorenzo Maggi*
- Old and New Steroids influence in Muscular Dystrophies | *Michela Guglieri*
- A gene therapy approach in Pompe Disease | *Federico Mingozzi*

10.40 - 12.40 | **Symposium**

Update of Pharmacological Treatment of SMA

Section 1

Chairman: *Enrico Bertini*

- What we know, don't know, and suspect about SMN function | *Fabrizio Gardoni*
- Modulating the levels of SMN beyond neurons: possible implications for clinical practice | *Valeria Sansone*

Section 2

Chairman: *Eugenio Mercuri*

- New perspectives in SMA Type 1 | *Adele D'Amico*
- New perspectives in later onset SMA | *Giacomo Comi*

Section 3

Chairman: *Claudio Bruno*

- Gene therapy in clinical trials | *Eugenio Mercuri*
- Beyond clinical trials: the Italian experience | *Emilio Albamonte*

12.40 - 13.10 | **Hot Topics:**

Chairman: *Antonella Pini*

Neonatal screening in neuromuscular disorders - *Francesco Danilo Tiziano*
Early diagnosis of neuromuscular diseases - *Alice Donati*

14.00 - 15.15 | **ORAL COMMUNICATIONS 1:**

New treatments for SMA

Chairmen: *Serenella Servidei, Roberto Massa*

14.00 - 14.15 | **Clinical and demographic features of patients with SMA on treatment with risdiplam: the iSMAC experience.**

E. Albamonte, G. Coratti, F. Salmin, A. Zanolini, M. Pane, MC Pera, D. Leone, L. Antonaci, A. D'Amico, M. Catteruccia, E. Bertini, C. Bruno, N. Brolatti, S. Messina, M. Sframeli, R. Piras, E. Mercuri, VA Sansone (Milano; Roma; Genova; Messina)

14.15 - 14.30 | **Amifampridine safety and efficacy in ambulatory SMA Type 3 patients: a randomized, placebo-controlled, crossover, phase 2 trial**

S. Bonanno, R. Giossi, R. Zanin, V. Porcelli, G. Ingenito, Z. Stevic, S. Peric, L. Maggi (Milano; Belgrade, Serbia)

- 14.30 - 14.45 | FIREFISH Part 2: 24-month efficacy and safety of risdiplam in infants with Type 1 spinal muscular atrophy (SMA)**
B.T. Darras, R. Masson, M. Mazurkiewicz-Beldzinska, K. Rose, H. Xiong, E. Zanoteli, G. Baranello, C. Bruno, D. Vlodavets, A. Dodman, M. El-Khairi, M. Gerber, K. Gorni, H. Kletzl, R. S Scalco, L. Servais
 (Boston, MA, USA; Milan, Italy; Gdansk, Poland; Sydney, Australia; Beijing, China; São Paulo, Brazil; London, UK; Genoa, Italy; Moscow, Russia; Basel, Switzerland; Welwyn Garden City, UK; Basel, Switzerland; Oxford, UK; Liège, Belgium; Paris, France)
- 14.45 - 15.00 | Long-term nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3**
L. Maggi, L. Bello, S. Bonanno, A. Govoni, C. Caponnetto, S.G. Grisanti, L. Passamano, M. Grandis, G. Nicocia, F. Trojsi, F. Cerri, A. Gardani, B. Risi, G. Giulio, M. Ferraro, V. Bozzoni, L. Caumo, R. Piras, R. Tanel, E. Saccani, M. Meneri, V. Vacchiano, G. Ricci, G. Sorarù, E. D'Errico, M.A. Maioli, I. Tramacere, S. Bortolani, G. Pavesi, R. Zanin, M. Silvestrini, L. Politano, A. Schenone, S. C. Previtali, A. Berardinelli, M. Turri, L. Verriello, M. Coccia, R. Mantegazza, R. Liguori, M. Filosto, G. Marrosu, G. Siciliano, I. L. Simone, T. Mongini, G.P. Comi, E. Pegoraro
 (Milano; Padova; Pisa; Genova; Napoli; Brescia; Torino; Carbonia; Trento; Parma; Bologna; Bari; Cagliari; Ancona; Bolzano)
- 15.00 - 15.15 | Onasemnogene abeparvovec palliative care experiences and challenges in type 1 Spinal Muscular Atrophy: the oldest care for the newest drug?**
A. Mercante, G. Perilongo, E. Salamon, A. Santini, F. Benini, C. Agosto (Padova)
- 15.15 - 17.00 | Poster session (Sessione non accreditata ECM)**
CHANNEL 1 - Dystrophinopathies and muscular dystrophies
 Chairmen: *Luca Bello, Adele D'Amico*
- CHANNEL 2 - Myasthenia, inflammatory myopathies, channelopathies**
 Chairmen: *Carmelo Rodolico – Matteo Garibaldi*
- CHANNEL 3 - Myopathies**
 Chairmen: *Enzo Ricci, Lorenzo Maggi*
- CHANNEL 4 - SMA and motor neuron disorders**
 Chairmen: *Antonio Di Muzio, Tiziana Mongini*
- 17.00 - 18.45 | ORAL COMMUNICATIONS 2**
Myopathies and muscular dystrophies: new phenotypes, biomarkers, and therapies – 1
 Chairmen: *Francesca Magri, Rita Barresi*
- 17.00 - 17.15 | Myalgia and fatigue as long term symptoms in COVID-19 patients: a 1-year follow up**
S. Cotti Piccinelli, A. Pilotto, V. Cristillo, F. Schiano di Cola, B. Risi, G. Bonzi, M. Mazzola, M. Filosto, A. Padovani (Brescia)

17.15 - 17.30 | Phenotypic spectrum of IGHMBP2 gene mutations and natural history study in a cohort of European patients

A. Govoni, A. Nuredini, M. Taiana, M. Nizzardo, A. D'Amico, E. Bertini, V. Sansone, E. Albamonte, S. Messina, F. Mari, E. Cesaroni, L. Porfiri, D. Tiziano, G.L. Vita, M. Sframeli, E. Merico, E. Schirinzi, G. Siciliano, J. A. F. Ramos, I. Ostrowska, M. Piontek, G.P. Comi, S. P. Corti
(Milano; Roma; Messina; Firenze; Ancona; Pisa; Cordoba, Spain; Szczecin, Poland; Zabrze, Poland)

17.30 - 17.45 | ASPIRO Gene Therapy Trial In X-Linked Myotubular Myopathy (XLMTM): Update on Preliminary Efficacy and Safety Findings

W. Miller, P. B. Shieh, N. Kuntz, J. J. Dowling, W. Müller-Felber, A. Blaschek, C. G. Bönnemann, A. R. Foley, D. N. Saade, A. M. Seferian, L. Servais, M. W. Lawlor, M. Noursalehi, S. Prasad, S. Rico
(San Francisco, CA, USA; Los Angeles, CA, USA; Chicago, IL, USA; Toronto, ON, Canada; Munich, Germany; Bethesda, MD, USA; Paris, France; Oxford, UK; Milwaukee, WI, USA)

17.45 - 18.00 | Safety, β -Sarcoglycan Expression, and Functional Outcomes From Systemic Gene Transfer of rAAVrh74.MHCK7.hSGCB in LGMD2E/R4

L. R. Rodino-Klapac, E. R. Pozsgai, S. Lewis, D. A. Griffin, A. S. Meadows, K. J. Lehman, K. Church, N. F. Reash, M. A. Iammarino, B. Powers, L. N. Alfano, L. P. Lowes, E. Koenig, S. Neuhaus, X. Li, L. Picaro, J. R. Mendell
(Cambridge, MA, USA; Columbus, OH, USA)

18.00 - 18.15 | An integrated and multiparametric system to identify diagnostic and prognostic parameters in neuromuscular diseases towards trials readiness.

G. Ricci, A. Tonacci, F. Torri, G. Aringhieri, F. Sansone, A. Rubegni, G. Siciliano, F. M. Santorelli, R. Conte and The InGene 2.0 project Consortium
(Pisa)

18.15 - 18.30 | Creatinine and CK plasma concentrations: possible role as biomarkers and correlation with age and muscular function in BMD.

P. Riguzzi, V. Zangaro, M. Villa, M. Cestarollo, E. Pegoraro, L. Bello (Padova)

18.30 - 18.45 | Titin truncating variants and their downstream effect on TTN transcripts

M. Savarese, M. Jobari, A. Vibola, H. Luque, TTN-RNAseq Working Group, P. Hackman, B. Udd (Folkhalsan Research Center)

Friday, 3rd December 2021

**08.00 - 08.40 | Early Symposium:
Myotubular Myopathy (MTM)**

Chairman: Angela Berardinelli

- MTM and natural history of the disease | Adele D'Amico
- Atypical clinical aspects of MTM | Cristina Molera

- 08.40 - 10.00 | Workshop 4 - Update on mitochondrial disease**
Chairman: *Michelangelo Mancuso*
- New emerging phenotypes | *Guido Primiano*
 - Biomarkers and outcome measures in mitochondrial disease: from diagnosis to clinical trials | *Costanza Lamperti*
 - Multi-omics approaches to improve mitochondrial disease diagnosis | *Dario Ronchi*
 - New therapeutic approaches: Small Molecules | *Michelangelo Mancuso*
- 10.00 - 10.30 | Invited Lecture**
Latest developments in Facioscapulohumeral Dystrophy
Introduction: *Rossella Tupler*
Guest: *Nicol Voermans*
- 10.30 - 11.30 | Workshop 5**
Inflammatory myopathies
Chairman: *Giovanni Antonini*
- Histopathological diagnosis | *Matteo Garibaldi*
 - Immune-mediated Necrotizing Myopathy | *Daniele Velardo*
 - Innovative treatments | *Massimiliano Mirabella*
- 11.30 - 12.00 | Invited Lecture “Recent update on dermatomyositis”**
Introduction: *Carlo Minetti*
Guest: *Ichizo Nishino*
- 12.00 - 13.00 | Symposium**
Patient assessment in view of new treatments for Pompe disease
Chairmen: *Antonio Toscano*
- Glycogen storage disease | *Tiziana Mongini*
 - Pre-symptomatic patients | *Olimpia Musumeci*
 - Respiratory disorders | *Grazia Crescimanno*
- 14.00 - 15.30 | ORAL COMMUNICATIONS 3**
Myopathies and muscular dystrophies: new phenomic, genomic, and pathogenetic aspects
Chairmen: *Grazia D'Angelo, Paola Tonin*
- 14.00 - 14.15 | Genotype-related respiratory progression in Duchenne Muscular Dystrophy – multicentre international study**
F. Trucco, D. Ridout, K. Maresb, M. Cbessbyre, P. Munot, A. Sarkozy, S. Robb, R. Quinlivan, M. Riley, C. Wallis, E. Chan., F. Abel, S. De Lucia, J.-Y. Hogrel, E. H. Niks, I. de Groot, L. Servais, V. Straub, V. Ricotti, A. Manzur, F. Muntoni (London, Uk; Paris, France; Leiden, The Netherlands; Liège, Belgium; Oxford, UK; Newcastle Upon Tyne, UK)

- 14.15 - 14.30 | Five years next-generation sequencing approach to neuromuscular symptoms and the prevalence of COL VI variants**
G. Marinella, B. Buchignani, A. Rubegni, G. Astrea, D. Cassandrini, MA Donati, M. Filosto, S. Gallone, F. Giannini, D. Lopercolo, MA Maioli, F. Magri, A. Malandrini, P. Mandich, F. Mari, R. Massa, S. Matà, M. Moggio, TE Mongini, E. Pegoraro, F. Ricci, G. Ricci, C. Rodolico, G. Siciliano, M. Sperti, C. Ticci, P. Tonin, FM Santorelli, R. Battini (Pisa; Firenze; Brescia; Torino; Siena; Cagliari; Milano; Genova; Roma; Padova; Messina; Verona)
- 14.30 - 14.45 | Improved use of the CPMS platform through tailored online training: the Italian EURO-NMD experience**
F. Bianchi, F. Fortunato, F. Giannini, A. Malandrini, V. Silan, N. Ticozzi, S. Fenu, S. Bonanno, C. Peduto, P. D'Ambrosio, G. Primiano, C. Sancricca, M. Sciacco, R. Brusa, M. Filosto, S. Cotti Piccinelli, E. Pegoraro, L. Solero, G. Gadaleta, C. Brusa, M. Cateruccia, D. Diodado, A. Pugliese, G. Nicocia, A. Ferlini, G. Siciliano (Pisa; Ferrara; Siena; Milano; Napoli; Roma; Brescia; Padova; Torino; Messina)
- 14.45 - 15.00 | Unveiling the relation between autophagy and Pompe disease**
F. M. Monasta, F. Blasevich, R. Mantegazza, L. Maggi, C. Bragato (Milano)
- 15.00 - 15.15 | Is fat detrimental for bone health in DMD? Insights from a longitudinal study.**
C. Panicucci, N. Brolatti, M. Pedemonte, E. Casalini, C. Minetti, M. Maghnie, C. Bruno, N. Di Iorgi (Genova)
- 15.15 - 15.30 | Preliminary Genome-Wide Association Study for identification and characterization of genetic modifiers of Duchenne muscular dystrophy**
D. Sabbatini, S. Vianello, A. Fusto, B. Merlo, A. Berardinelli, S. Parravicini, C. Bruno, C. Panicucci, G. P. Comi, F. Magri, A. D'Amico, M. Catteruccia, L. Travagliani, Grazia D'Angelo, V. Sansone, A. Di Bari, T. Mongini, C. Brusa, L. Maggi, E. Canioni, A. Gallone, M. Pane, D. Leone, L. Politano, E. Picillo, V. Nigro, S. Messina, G. Vita, G. Sorarù, L. Bello, E. Pegoraro (Padova; Genova; Milano; Roma; Bosisio Parini (Lecco; Torino; Napoli; Messina)
- 15.30 - 17.15 | ORAL COMMUNICATIONS 4**
Natural history and new diagnostic tools in neuromuscular disorders
Chairman: Giorgio Tasca, Sabrina Ravaglia
- 15.30 - 15.45 | Limb girdle muscular dystrophy type R1/2A and R2/2B: three decades of natural history**
A. LoMauro, E. Diella, A. Russo, M. Delle Fave, C. Pistininzi, E. Marchi, R. Pascuzzo, S. Vantini, A. Aliverti, M.G. D'Angelo (Milano)
- 15.45 - 16.00 | Comorbidities in Myasthenia Gravis: a report from 178 patients**
S. Iacono, V. Di Stefano, A. Lupica, A. Gagliardo, P. Lanza, M.G. Rispoli, L. Ferri, F. Brighina, A. Di Muzio (Palermo; Chieti)

- 16.00 - 16.15 | A clinical follow up study in a cohort of Italian children affected by LMNA gene mutations.**
B. Buchignani, G. Marinella, A. Rubegni, D. Cassandrini, S. Frosini, F.M. Santorelli, G. Astrea, R. Battini (Pisa)
- 16.15 - 16.30 | Congenital myasthenic syndrome: natural history of an Italian cohort of patients**
A. Gallone, A. Pugliese, S. Bonanno, M. Garibaldi, C. Rodolico, L. Maggi (Milano; Messina; Roma)
- 16.30 - 16.45 | Refractory Myasthenia Gravis: characteristics of an Italian cohort of patients**
G. Nicocia, A. Pugliese, R. Frangiamore, S. Bonanno, A. Gallone, C. Antozzi, F. Baggi, C. Rodolico, R. Mantegazza, L. Maggi (Milano; Messina)
- 16.45 - 17.00 | Linked-read WGS as further step to study unsolved NMD cases**
M.E. Onore, A. Torella, F. Musacchia, F. Del Vecchio Blanco, P. D'Ambrosio, M. Zanobio, G. Piluso, V. Nigro (Napoli; Pozzuoli)
- 17.00 - 17.15 | Immunofluorescence signal intensity measurements as a semi-quantitative tool to assess sarcoglycan complex expression in muscle biopsy**
F. Poggetti, F. Magri, M. Ripolone, M. Sciacco, M. Moggio, G.P. Comi, S. Zanotti (Milano)
- 17.15 - 18.45 | ORAL COMMUNICATIONS 5**
Myopathies and muscular dystrophies: new phenotypes, biomarkers, and therapies – 2
Chairmen: Giovanni Meola, Monica Sciacco
- 17.15 - 17.30 | Medaka fish as a new model to investigate skeletal muscle performance**
M. Marcello, I. Morotti, M. Caremani, M. Savarese, B. Udd, I. Conte, V. Nigro, M. Linari (Firenze; Helsinki, Finland; Pozzuoli; Napoli)
- 17.30 - 17.45 | Characterization of DNA methylation status of the D4Z4 locus as epigenetic biomarker for the molecular diagnosis of FSHD**
V. Caputo, D. Megalizzi, C. Bax, M. Ranieri, L. Colantoni, G. Tasca, E. Ricci, C. Caltagirone, R. Cascella, E. Giardina, C. Strafella (Roma)
- 17.45 - 18.00 | AIGkit, the mobile app for patients with Pompe disease: results and future prospectives**
E. Lai, G. Ricci, E. Schirinzi, G. Siciliano (Pisa)
- 18.00 - 18.15 | Selective P2X7 antagonism ameliorates the dystrophic phenotype of a-sarcoglycan-deficient mice by dampening muscle inflammation and fibrosis**
L. Raffaghello, E. Principi, S. Baratto, C. Panicucci, S. Pintus, F. Antonini, G. Del Zotto, S. Bruzzone, P. Scudieri, C. Minetti, E. Gazzero, C. Bruno (Genova; Berlin, Germany)

- 18.15 - 18.30 | **Antisense oligonucleotide (ASO)-based in vitro modulation of GAA expression in Late-Onset Pompe Disease**
D. Ronchi, S. Lucchiari, F. Magri, M. Garbellini, S. Salani, S. Zanotti, P. Ciscato, M. Sciacco, M. Moggio, S. Corti, N. Bresolin, GP Comi (Milano)
- 18.30 - 18.45 | **Solve unsolved neuromuscular cases: collaborative reanalysis of WES data through the Solve-RD Genome-Phenome Analysis Platform (GPAP).**
A. Torella, R. Zeuli, A. Varavallo, F. Del Vecchio Blanco, C. Angelini, N. Brunetti Pierri, A. Wischmeijer, Solve-RD DITF-EuroNMD, G. Piluso, V. Nigro (Napoli; Telethon Institute of Genetics and Medicine (TIGEM); Padova; Bolzano)

18.45 | **Assemblea Soci AIM**

Saturday, 4th December 2021

- 08.30 - 09.30 | **Wake up Muscle Club**
Chairmen: *Chiara Fiorillo, Giulia Ricci*
- 08.30 - 08.42 | **"Sudden weakness in a 56-year-old Asian male"**
B. Labella, D. Pezzini, A. Costa, L. Poli, M. Magoni, A. Padovani (Brescia)
- 08.42 - 08.54 | **"A case of severe sensorimotor polyneuropathy and distal myopathy associated with vocal cord and pharyngeal weakness"**
A. Manini, D. Velardo, P. Ciscato, C. Cinnante, F. Magri, M. Moggio, GP Comi, S. Corti, D. Ronchi (Milano)
- 08.54 - 09.06 | **"Reorientation of muscle biopsy as a diagnostic hint"**
M. Meznaric, J. Zidar (Ljubljana, Slovenia)
- 09.06 - 09.18 | **"A case of exercise intolerance and deafness"**
C. Panicucci, M. Traverso, A. Pini, M. Giannotta, M.L. Valentino, M. Scala, M. Pedemonte, C. Minetti, F. Zara, C. Fiorillo, C. Bruno (Genova; Bologna)
- 09.18 - 09.30 | **"Cognitive impairment and slowly progressive muscular weakness in a 6-year-old boy"**
S. Antognozzi, F. Magri, P. Ciscato, L. Napoli, D. Velardo, G. Scuvera, A. Giacobbe, D. Milani, M. Sciacco, M. Moggio, S. Corti, N. Bresolin, GP Comi, D. Ronchi (Milano)
- 09.30 - 11.00 | **Workshop 6**
Next generation molecular diagnostics
Chairmen: *Vincenzo Nigro, Filippo Santorelli*
 • EXPEDITing in vivo gene therapy with AAV vectors | *Alberto Auricchio*

- How to integrate genomic and exomic data in clinical practice | *Alessandra Ferlini*
- The risk of false positives and false negatives in genomic data; The SOLVE RD experience | *Steven Laurie*
- Genetic approach to undiagnosed myopathies | *Vincenzo Nigro*

11.00 - 12.30 | AIM meets Patients and Family Associations

Chairmen: *Carlo Minetti, Gabriele Siciliano, Luisa Politano*

- UILDM 60 years of activity - *Marco Rasconi*
- Telethon Registries - *Anna Ambrosini*
- Voice to the Associations (*AIDMED, AIG, AMAR, AISED, Altro Domani, ASAMSI, Ass Dodò, AICA3, COL6, Di.MIO, Famiglie SMA, FMM,FSHD It, GFB, Mitocon, Parent Project, UILDM, and others*)

12.30 | Awards Ceremony for the best works

13.00 | Conclusions



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POSTER SESSION

Poster Session

Thursday, 2nd December 2021

15.15 - 17.00 | POSTER – CHANNEL 1

Dystrophinopathies and muscular dystrophies

Chairmen: *Luca Bello, Adele D'Amico*

15.15 - 15.23 | [Care for adults with Duchenne Muscular Dystrophy \(DMD\): a single centre experience.](#)

C. Brusa, E. Rolle, F. Rossi, I. Cavallina, R. D'Alessandro, G. Gadaleta, G. Urbano, F. Ricci, T. Mongini (Torino)

15.23 - 15.31 | [Becker Muscular Dystrophy and 7q11.23 microduplication syndrome in a boy with developmental delay, bicuspid aorta and interatrial septum defect](#)

E. Picillo, P. D'Ambrosio, L. Passamano, A. Torella, V. Nigro (Napoli; Messina; Pozzuoli)

15.31 - 15.39 | [Cerebral alteration patterns in children with Duchenne Muscular Dystrophy: a machine learning approach on magnetic resonance images](#)

D. Peruzzo, T. Ciceri, S. Mascheretti, V. Lampis, F. Arrigoni, A. Giubergia, F. M. Villa, A. Crippa, M. Nobile, E. Mani, A. Russo, M. G. D'Angelo (Bosisio Parini)

15.39 - 15.47 | [Detection of dystrophin isoforms transcripts in human adult control brain using in-situ RNA analysis](#)

M.S. Falzarano, M. Mietto, R. Rossi, F. Fortunato, R. Selvatici, M. Gessi, F. Montanaro, J. Morgan, F. Muntoni, A. Ferlini (Ferrara; London, UK; Roma)

15.47 - 15.55 | [A novel DMD gene splice-site variant in an Italian boy with Becker muscular dystrophy: a clinical and genetic study](#)

G. Lanzi, B. Risi, S. Cotti Piccinelli, A. Galvagni, C. Romani, C. Fiorillo, M. Traverso, C. Bruno, A. Padovani, M. Filosto (Brescia; Genova)

15.55 - 16.03 | [Delay in Duchenne Muscular Dystrophy Progression with Eteplirsen: Longer Time to Loss of Ambulation Versus Standard of Care](#)

J. Iff, G. Bungey, A. Paine, B. Han, H. Gordish-Dressman, E. Henricson, L. Picaro, C. McDonald (Cambridge, Massachusetts, USA; London, UK; Washington DC, USA; California, USA)

16.03 - 16.11 | [Disease characterization of ambulant patients with Becker muscular dystrophy: histopathological, functional and imaging data from Givinstat trial cohort](#)

D. Velardo, M. Ripolone, S. Zanotti, F. Magri, C. M. Cinnante, S. Montrasio, S. Cazzaniga, E. H. Niks, M. Sciacco, P. U. Bettica, G. P. Comi (Milano; Leiden, the Netherlands)

16.11 - 16.19 | [Evaluation of 6MWT as functional measure in Becker muscular dystrophy](#)

V. Zangaro, L. Bello, P. Riguzzi, M. Villa, S. Mastellaro, M. Cestarollo, E. Pegoraro (Padova)

16.19 - 16.27 | [An integrative splicing predictor pipeline to prioritise DMD deep intronic variants](#)

R. Zeuli, M. Zanobio, M.E. Onore, F. Romano, M.M. Napoli, E. Picillo, A. Torella, F. Del Vecchio Blanco, G. Piluso, V. Nigro (Napoli; Pozzuoli)

16.27 - 16.35 | [Antisense morpholino-based in vitro correction of a novel pseudoexon-generating mutation in the SGCB gene](#)

F. Magri, S. Salani, F. Fortunato, S. Zanotti, P. Ciscato, S. Gerevini, L. Maggi, M. Sciacco, M. Moggio, S. Corti, N. Bresolin, D. Ronchi, GP Comi
(Milano; Bergamo)

16.35 - 16.43 | [Two cases of autosomal dominant Ullrich congenital muscular dystrophy due the same de novo mutation in the COL6A3 gene](#)

E. Picillo, L. Passamano, A. Torella, F. Del Vecchio Blanco, M.E. Onore, M. Zanolio, R. Zeuli, L. Politano, A. Selicorni, V. Nigro (Napoli; Pozzuoli; Como)

16.43 - 16.51 | [Oculo-pharyngeal muscular dystrophy: phenotypic and genotypic studies in Abruzzo](#)

M.G. Rispoli, L. Ferri, P. Ajdinaj, A. Di Muzio (Chieti-Pescara; Chieti)

15.15 - 17.00 | **POSTER - CHANNEL 2**

Myasthenia, inflammatory myopathies, channelopathies

Chairmen: Carmelo Rodolico – Matteo Garibaldi

15.15- 15. 23 | [Characterization of p.N1180I sodium channel mutant associated to myotonia and myopathy in Italian patients](#)

C. Altamura, A. Farinato, C. Campanale, M. R. Carratù, J.-F. Desaphy (Bari)

15.23 - 15. 31 | [Mutations associated with hypokalemic periodic paralysis: from hotspot regions to complete analysis of CACNA1S and SCN4A genes.](#)

R. Brugnoli, E. Canioni, M. Filosto, A. Pini, P. Tonin, T. Rossi, C. Canavese, M. Eoli, G. Siciliano, G. Lauria, R. Mantegazza, L. Maggi
(Milano; Brescia; Verona; Ancona; Torino; Pisa)

15.31 - 15. 39 | [SARS-CoV-2 infection in myasthenic patients from Liguria](#)

E. Scarsi, S. Grisanti, A. Cella, E. Narciso, C. Cabona, A. Beronio, A. Assini, M. Del Sette, F. Bandini, L. Benedetti, V. Prada, A. Schenone, M. Grandis (Genova; La Spezia; Iowa City, IA, USA)

15.39 - 15. 47 | [COVID-19 vaccine and dermatomyositis: is there an association?](#)

P. Ajdinaj, L. Ferri, M. G. Rispoli, F. Barbone, M. A. De Rosa, D. Angelucci, G. Andreassi, M. Amatetti, P. Amerio, A. Di Muzio (Chieti-Pescara; Ortona)

15.47 - 15. 55 | [A very late onset AChR and MuSK double positive myasthenia gravis](#)

A. Pugliese, P. D'Ambrosio, G. Nicocia, S. Messina, A. Toscano, C. Rodolico (Messina)

15.55 - 16. 03 | [Congenital myasthenia associated with new variant of the PREPL gene](#)

M. Traverso, M. Scala, S. Baratto, M. Di Duca, C. Campana, M. Cataldi, C. Panicucci, M. Pedemonte, C. Bruno, C. Minetti, F. Zara, C. Fiorillo (Genova)

16.03 - 16.11 | [Rituximab in Refractory Myasthenia Gravis: what we learned after 10 years. A single centre experience.](#)

G. Greco, E. Frezza, M. Goglia, L. Boffa, R. Massa (Roma)

16.11 - 16.19 | Impact of COVID-19 and vaccines in myasthenia gravis: experience from an Italian cohort

A. Lupica, V. Di Stefano, S. Iacono, A. Pignolo, M. Quartana, A. Gagliardo, B. Fierro, F. Brighina (Palermo)

16.19 - 16.27 | Mutation of LRSAM1 gene: a novel cause of congenital myasthenic syndrome.

A. Pugliese, G. Nicocia, P. D'Ambrosio, S.A. Musumeci, A. Toscano, C. Rodolico (Messina; Troina)

15.15 - 17.00 | POSTER – CHANNEL 3

Myopathies

Chairmen: Enzo Ricci, Lorenzo Maggi

15.15 - 15.23 | Severe progressive course in an NLSDM patient put on MCT dietary treatment

C. Angelini, S. Missaglia, D. Tavian (Padova; Milano)

15.23 - 15.31 | The role of muscle biopsy: A 15 years single center experience.

S. Cotti Piccinelli, E. Baldelli, B. Risi, F. Caria, A. Padovani, M. Filosto (Brescia; Fano)

15.31 - 15.39 | Respiratory function in Myotonic Dystrophy Type 1: a Retrospective Study

C. R. Ferrari Aggradi, E. Falcier, A. Lizio, A. Pirola, J. Casiraghi, A. Zanolini, E. Carraro, L. Mauro, F. Rao, E. Roma, A. Iannello, E. De Mattia, A. Barp, S. Lupone, V. Gatti, C. Italiano, V. A Sansone (Milano)

15.39 - 15.47 | Establishing Biomarkers and Clinical Endpoints in Myotonic Dystrophy Type 1 (END-DM1): Italian experience

M.C. Frisoni, C. Ferrari-Aggradi, L. Mauro, A. Di Bari, J. Refran, S. Becchiati, F. Iossa, A. Zanolini, K. Eichinger, J. Dekdebrun, J. St Romain, N. Johnson, C. Thornton, VA Sansone (Milano; Rochester, New York; Richmond, VA)

15.47 - 15.55 | Cardiac magnetic resonance findings and outcome in type 1 myotonic dystrophy

M. Leali, A. Aimo, G. Ricci, G. Todiere, G. Vergaro, C. Grigoratos, A. Giannoni, F. Torri, F. Baldinotti, G. Donato Aquaro, G. Siciliano, M. Emdin, C. Passino, A. Barison (Pisa)

15.55 - 16.03 | A novel distal vacuolar myopathy caused by a large expansion of the PLIN4 gene- clinical, histological and imaging data.

L. Maggi, S. Gibertini, E. Iannibelli, A. Gallone, C. Bragato, S. Bonanno, F. Blasevich, R. Mantegazza, M. Mora, A. Ruggieri (Milano)

16.03 - 16.11 | Prevalent muscle involvement in two siblings with Glycogen Storage Disease type III

L. Passamano, M. Savarese, A. Torella, M. Scutifero, E. Picillo, A. Palladino, V. Nigro, L. Politano (Napoli; Helsinki, Finland; Pozzuoli)

16.11 - 16.19 | [“Natural history of skeletal muscle laminopathies: a 2-year prospective study”](#)

L.S. Santovito, S. Bonanno, B. Pasanisi, A. Gallone, F. Ricci, I. Tramacere, R. Zanin, S.C. Previtali, L. Maggi (Chicago, IL, USA; Milano; Torino)

16.19 - 16.27 | [Muscle MRI images in patients with Chronic Progressive External Ophthalmoplegia](#)

M. Sperti, C. Nesti, M. Bartolini, F.M. Santorelli, S. Matà (Firenze; Pisa)

16.27 - 16.35 | [Psychopathology and coping styles in patients with neuromuscular disorders: a follow-up study during the COVID pandemic](#)

M. Nobile, A. Tarabelloni, F. Tizzoni, P. Colombo, A. Tesei, A. Russo, M. Molteni, S. Previtali, Y. Falzone, A. Berardinelli, M.G. D'Angelo (Bosisio Parini; Milano; Pavia)

16.35 - 16.43 | [Late-onset diffuse myalgias and multiple subarachnoid cysts. A case report](#)

F. Torri, L. Chico, F. Baldinotti, M.A. Caligo, M.R. D'Apice, G. Ricci, G. Siciliano (Pisa)

15.15 - 17.00 | POSTER – CHANNEL 4

SMA and motor neuron disorders

Chairmen: Antonio Di Muzio, Tiziana Mongini

15.15 - 15.23 | [Dysregulation of lncRNAs and mRNA targets during skeletal muscle development in a patient-derived induced pluripotent stem cell \(iPSC\) model of Amyotrophic Lateral Sclerosis.](#)

E. Giagnorio, C. Malacarne, E. Salvi, P. Bossolasco, D. Bardelli, A. Ratti, G. Lauria, R. Mantegazza, S. Bonanno, S. Marcuzzo (Milano; Monza)

15.23 - 15.31 | [Insights for the development of a miRNA-based therapeutic strategy in ALS exploiting iPSC-derived motor neurons and exosomes.](#)

V. Melzi, M. Rizzuti, D. Gagliardi, M. Meneri, P. Masrori, F. Biella, P. Van Damme, GP Comi, M. Nizzardo, S. Corti (Milano; Leuven, Belgium)

15.31 - 15.39 | [Quantitative MRI evaluation of muscle involvement of lower limbs in amyotrophic lateral sclerosis \(ALS\): a pilot longitudinal study](#)

M. Paoletti, L. Diamanti, S.I. Muzic, E. Ballante, F. Solazzo, L. Foppoli, X. Deligianni, S. Santini, S. Figini, N. Bergsland, A. Pichiechio (Pavia; Basel, CH; Buffalo, NY, United States; Milano)

15.39 - 15.47 | [Parental mosaicism in AARS1 – related hereditary neuropathy](#)

M. Neri, E. Sette, F. Fortunato, C. TrabANELLI, A. Margutti, P. Rimessi, M. Fabris, V. Tugnoli, F. Gualandi, R. Selvatici, A. Ferlini (Ferrara)

15.47 - 15.55 | [Use of 3d printer in a rehab lab for the creation of customized assistive devices with users with radiation-induced brachial plexopathy to increase participation: case study](#)

F. Pilla, P.G. Ianes, S. Casagrande, B. Gasperini, R. Zuccarino (Trento)

15.55 - 16.03 | Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen

S. Bonanno, R. Zanin, L. Bello, I. Tramacere, V. Bozzoni, L. Caumo, M. Ferraro, S. Bortolani, G. Sorarù, M. Silvestrini, V. Vacchiano, M. Turri, R. Tanel, R. Liguori, M. Coccia, R. E. Mantegazza, T. Mongini, E. Pegoraro, L. Maggi (Milano; Padova; Torino; Ancona; Bologna; Bolzano; Trento)

16.03 - 16.11 | Exploring face mobility in SMA

E. Carraro, E. Pegolo, F. Cibir, MC Frisoni, F. Salmin, S. Becchiati, Z. Sawacha, VA Sansone (Milano; Padova)

16.11 - 16.19 | 6MWT as measure of fatigability in sma type 3 patients treated with nusinersen

A. Govoni, G. Ricci, M. Meneri, S. Bonanno, L. Bello, C. Caponnetto, L. Passamano, M. Grandis, F. Trojsi, F. Cerri, G. Gadaleta, V. Bozzoni, L. Caumo, R. Tanel, E. Saccani, V. Vacchiano, G. Sorarù, E. D'Errico, I. Tramacere, S. Bortolani, G. Pavesi, R. Zanin, M. Silvestrini, L. Politano, A. Schenone, S.C. Previtali, A. Berardinelli, M. Turri, L. Verriello, M. Coccia, R. Mantegazza, R. Liguori, M. Filosto, M. Maioli, G. Marrosu, I.L. Simone, T. Mongini, S. Corti, E. Pegoraro, G. Siciliano, G. Comi, L. Maggi (Milano; Pisa; Genova; Napoli; Torino; Parma; Bologna; Bari; Ancona; Pavia; Bolzano; Udine; Brescia; Cagliari)

16.19 - 16.27 | Neurofilament light chain and Profilin-1 in adult SMA patients under nusinersen treatment: 26-months follow up

G. Musso, V. Bozzoni, L. Caumo, L. Bello, C. Cosma, G. Sorarù, M. Plebani, E. Pegoraro (Padova)

16.27 - 16.35 | Cell-penetrating peptides-conjugated Morpholino: a novel therapeutic approach for the treatment of Spinal Muscular Atrophy symptomatic cases

M. Rizzuti, M. Bersani, E. Pagliari, M. Garbellini, D. Saccomanno, N. Bresolin, G. P. Comi, S. Corti, M. Nizzardo (Milano)

16.35 - 16.43 | Oro-facial and bulbar involvement in SMA (OBI-SMA): outcome measures & end-point assessments

F. Salmin, C. Cattaneo, J. Lops, E. Carraro, G. Coratti, MC Pera, A. Pirola, A. Di Bari, E. Albamonte, A. Zanolini, G. Palazzo, L. Mauro, M. Pane, E. Mercuri, VA Sansone (Milano; Roma)

16.43 - 16.51 | The experience with Nusinersen (Spinraza) in pediatric SMA patients in Emilia-Romagna in a regional network

G. Scarpini, M. Giannotta, R. Not, P. Capelli, C. Testoni, F. Pisani, B. Piccolo, C. Fusco, D. Frattini, G. Vergine, M. Farina, J. Savajlia, F. Marchetti, G. Piccinini, A. Pini (Bologna; Parma; Reggio Emilia; Rimini; Romagna; Ravenna)

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Psicologo:	<i>Psicoterapia; Psicologia;</i>
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