ASSOCIAZIONE ITALIANA
MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

XXIII NATIONAL CONGRESS
OF ITALIAN ASSOCIATION of MYOLOGY

PADOVA
8th - 10th June 2023

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XXIII NATIONAL CONGRESS
OF ITALIAN ASSOCIATION
of MYOLOGY

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PROGRAM

ITALIAN ASSOCIATION of MYOLOGY

Thursday, 8th June 2023

MANTEGNA HALL

10.10 – 10.40 | LECTURE NO CME

Challenges delivering ERT in lysosomal disorders: special considerations in LOPD
Brian Fox (New York, USA) introduced by *Corrado Angelini (Padova)*

10:45 – 11:00 GREETINGS AND INTRODUCTION
Chairpersons: *Giacomo Comi (Milano), Maurizio Corbetta (Padova), Elena Pegoraro (Padova)*

11.00 – 11.30 | MAIN LECTURE

Translating knowledge on hereditary neuromuscular childhood diseases into therapies
Francesco Muntoni (Londra, UK) introduced by *Giacomo Comi (Milano)*

11.30 – 13.00 | WORKSHOP
SLEEP DISORDERS IN NEUROMUSCULAR DISEASES: TREATABLE CONDITIONS
Chairpersons: *Claudio Cherchi (Roma), Antonio Trabacca (Brindisi)*

11.30 | Sleep disorders in neurological and neuromuscular conditions:
diagnostic tools and treatment approach | *Michelangelo Maestri (Pisa)*

11.50 | Sleep and Respiratory Disorders in Duchenne Muscular Dystrophy
Maria Grazia D'Angelo (Milano)

12.10 | Disorders of sleep in LOPD and response to ERT | *Sabrina Ravaglia (Pavia)*

12.30 | Sleep Disorders in SMA | *Elisabetta Verrillo (Roma)*

12.50 | Discussion

13.00 – 14.00 | Lunch

13.00 – 15.00 | Poster Viewing

14.00 – 15.00 | SYMPOSIUM NO CME

New frontiers in SMA
Chairperson: *Giacomo Comi (Milano)*

14.00 | What is next for SMA Management? | *Stefania Corti (Milano), Eugenio Mercuri (Roma)*

14.30 | How technology innovation can support the multidisciplinary approach
Davide Korn (Roma)

15.00 – 16.00 | INVITED TALKS
Chairpersons: *Alessandra Ferlini (Bologna), Rossella Tupler (Modena)*

15.00 | Preimplantation Genetic Test (PGT) | *Daniela Zuccarello (Padova)*

15.30 | Mechanisms regulating skeletal muscle growth and atrophy
Marco Sandri (Padova)

16.00 – 17.00 | New Therapeutic Approaches in Pompe Disease
Chairpersons: *Alberto Burlina (Padova), Gabriele Sicialiano (Pisa)*

16:00 | Novelties in the treatment of infantile Pompe disease | *Serena Gasperini (Monza)*

16.30 | Long term clinical study data on late onset Pompe disease
Olimpia Musumeci (Messina)

17.00 – 17.30 | Coffee Break

17.30 – 18.30 | WORKSHOP | GENDER ISSUES IN NEUROMUSCULAR DISORDERS
Chairpersons: *Chiara Fiorillo (Genova), Paola Tonin (Verona)*

17.30 | Gender issues in SMA | *Luca Bello (Padova)*

17.50 | FSHD different courses in males and females | *Massimiliano Filosto (Brescia)*

18.10 | New avenues to treat androgen receptor' in Kennedy disease| *Maria Pennuto (Padova)*

18.30 – 19.00 | LECTURE NO CME

Newborn screening for SMA: the new scenarios
Federica Ricci (Torino)

19.00 - 20.00 | SYMPOSIUM NO CME

The Motor Pool in SMA
Chairperson: *Elena Pegoraro (Padova)*

19:00 | Welcome and Introduction | *Elena Pegoraro (Padova)*

19:05 | An introduction to motor pools and neurodegeneration | *Sonia Messina (Messina)*

19:20 | Tracing the motor pool-specific vulnerabilities | *Toby Ferguson (Boston, USA)*

19:35 | Clinical implication & clinical case | *Valeria Sansone (Milano)*

19:50 | Questions and Answers

20.00 | Welcome Cocktail

PETRARCA HALL

11.30 - 13.00 | ORAL COMMUNICATIONS | SMA
Chairpersons: *Adele D'Amico (Roma), Claudio Bruno (Genova)*

11.30 | Search for new CSF biomarkers in SMA patients treated with Nusinersen
C. Panicucci, M. Bartolucci, E. Sahin, R. Russo, M. Valletta, N. Brolatti, M. Pedemonte, S. Casalini, S. Baratto, A. D'Amico, M. Pane, M. Sframeli, C. Stancanelli, S. Messina, E. Albamonte, V. Sansone, E. Mercuri, A. Chambery, U. Sezerman, A. Petretto, C. Bruno.
Genova, Istanbul, Caserta, Roma, Messina, Milano

11.45 | Pregnancy experience in women with spinal muscular atrophy: an overview in the cohort of the University of Naples Federico II
R. Piera Bencivenga, D. Zoppi, A. Russo, E. Cassano, S. Tozza, R. Iodice, R. Dubbioso, F. Manganelli, L. Ruggiero.
Napoli

12.00 | Patients and caregivers expectations on possible functional changes following disease-modifying treatment in spinal muscular atrophy
M. C. Pera, G. Coratti, J. L. Casiraghi, C. Bravetti, A. Fedeli, M. Strika, E. Albamonte, L. Antonaci, D. Rossi, M. Pane, V. Sansone, E. Mercuri.
Roma

12.15 | Swallowing and feeding in Spinal Muscular Atrophy (SMA) type 1: the SFERA project and the path towards a diagnostic and treatment algorithm
S. Gandolfi, C. Dosi, M.R. Scopelliti, E. Minacapilli, R. De Amicis, S. Bertoli, A. Mandelli, A. Campari, A. Schindler, R. Masson, on behalf of the SFERA working group.
Milano

12.30 | Facial nerve vulnerability in spinal muscular atrophy and motor unit number index (MUNIX) of the orbicularis oculi muscle
A. Barp, E. Carraro, F. Salmin, A. Lizio, M. Cheli, V. Sansone.
Milano, Valsugana

12.45 | Molecular and biochemical characterization of patient-derived cellular models for Spinal Muscular Atrophy with Myoclonus Epilepsy (SMA-PME)
D. Ronchi, S. Salani, M. Garbellini, M. S. Hidalgo Moreno, S. Antognozzi, M. Aureli, S. Corti, G. P. Comi.
Milano

13.00 – 14.00 | Lunch

13.00 – 15:00 | Poster Viewing

17:30 - 18:30 | ORAL COMMUNICATIONS | DIAGNOSTIC AND THERAPEUTIC TOOLS
Chairpersons: *Marco Savarese (Helsinki, Finland), Giulia Ricci (Pisa)*

17:30 | Engineering human stem cells for advanced neuromuscular disease and therapy modelling
V. M. Lionello, L. Pinton, S. Dastidar, D. Moore, S. Choi, H. Steele-Stallard, S. Jalal, Y. Jiang, P. Zammit, F. S. Tedesco.
Londra

17:45 | Solving the NGS negative cases: improved WES protocols to make increasingly accurate genetic diagnoses
A. Torella, M. E. Onore, E. Picillo, A. Budillon, G. Piluso, L. Politano, V. Nigro.
Napoli

18:00 | A comparison of different NGS-based strategies for the routine clinical diagnosis of neuromuscular disorders
C. Bertolin, F. Boaretto, S. Volta, E. Pegoraro, L. Bello, G. Sorarù, C. Viscomi, M. Zeviani, L. Salviati.
Padova

18:15 | OMICS analysis to identify the genetic causes of undiagnosed neuromuscular phenotypes: the UNIFE experience whthin the SOLVE-RD Project
R. Selvatici, M. Neri, F. Gualandi, S. Bigoni, F. Fortunato, E. Bertini, M. Pane, E. Mercuri, A. Ferlini.
Ferrara, Roma

PALLADIO HALL

17.30 - 18.30 | ORAL COMMUNICATIONS | DISEASE MECHANISMS AND PATHOGENESIS
Chairpersons: *Roberto Massa (Roma), Costanza Lamperti (Milano)*

17.30 | RNA-seq data analysis in RYR1related congenital myopathies with cores and minicores: an Italian population study
D. Sabbatini, A. Fusto, M. Suman, D. Gorgoglione, S. Vianello, C. Romualdi, C. Marchioretti, M. Pennuto, G. Szabadkai, L. Bello, E. Pegoraro.
Padova, Londra

17.45 | Medaka fish Oryzias latipes a promising model to study titinopathies: the HMERF case study
V. Cetrangolo, M. Savarese, I. Conte, L. Polishchuk, V. Nigro, M. Linari, B. Udd.
Pozzuoli

18.00 | Aberrant Adenosine Triphosphate Release and Impairment of P2Y2-Mediated Signaling in Sarcoglycanopathies
S. Baratto, A. Benzi, C. Astigiano, L. Sturla, C. Panicucci, K. Mamchaoi, L. Raffaghello, S. Bruzzone, E. Gazzerro, C. Bruno.
Genova, Berlino, Parigi

18.15 | Functional characterization suggests that ACTN2 frameshift variants cause distal myopathy through protein aggregation
J. Ranta-aho, P. H. Jonson, J. Sarparanta, B. Udd, M. Savarese.
Helsinki

Friday, 9th June 2023

MANTEGNA HALL

08:00 - 08:30 | MAIN LECTURE
Chairpersons: *Giacomo Pietro Comi (Milano), Luisa Politano (Napoli)*

08:00 | Cardiac involvement in female D/BMD-gene mutation carriers
John Bourke (Newcastle, UK)

08.30 – 10.00 | WORKSHOP | MITOCHONDRIAL DISORDERS
Chairpersons: *Michelangelo Mancuso (Pisa), Massimo Zeviani (Padova)*

08:30 | Cardiorespiratory involvement in primary mitochondrial diseases
Serenella Servidei (Roma)

08:50 | Mitochondrial optic neuropathies: differential diagnosis and therapeutic strategies
Chiara La Morgia (Bologna)

09:10 | Mitochondrial Myopathies: Models and therapies
Carlo Viscomi (Padova)

09:30 | SURF1 therapeutic development in non-human mammals
Dario Brunetti (Milano)

09:50 | Discussion

10.00 – 10.30 | NEREO BRESOLIN LECTURE ON MITOCHONDRIAL DISORDERS

A Journey to Develop Therapy for Thymidine Kinase 2 Deficiency
Michio Hirano (New York) introduced by Antonio Toscano (Messina)

10.30 – 11.00 | Coffee Break

11.00 – 12.30 | ORAL COMMUNICATIONS | LGMD AND FSHD
Chairpersons: *Francesca Magri (Milano), Giorgio Tasca (Roma)*

11.00 | Clinical and genetic analysis of the Italian cohort of LAMA2-Related Dystrophy (LAMA2-RD)
A. Zambon, S. Messina, M. Sframeli, A. Pini, M. L. Valentino, C. Fiorillo, C. Bruno, C. Panicucci, F. Ricci, T. Mongini, G. D'Angelo, R. Cima, G. Ricci, L. Politano, M. Pane, E. Mercuri, E. Pegoraro, L. Bello, E. Albamonte, F. Trucco, V. Sansone, A. Berardinelli, G. Astrea, R. Battini, M. Filosto, S. Cotti Piccinelli, F. Magri, D. Velardo, G. P. Comi, I. Moroni, F. Bruschi, L. Maggi, M. Catteruccia, E. Bertini, A. D'Amico, S. C. Previtali.
Milano, Messina, Genova, Torino, Pisa, Napoli, Roma, Padova, Brescia

11.15 | The role of the NAD+/CD38 axis in the pathophysiology of alfa-sarcoglycanopathy
A. Benzi, A. Amaro, S. Pintus, F. Antonini, F. Reggiani, S. Baratto, E. Principi, G. Del Zotto, D. Cassandrini, A. D'Amico, A. Malandrini, E. Malfatti, T. Mongini, E. Pegoraro, S. Previtali, C. Rodolico, G. Tasca, F. Morandi, F. Malavasi, U. Pfeffer, S. Bruzzone, C. Bruno, L. Raffaghello.
Genova, Pisa, Roma, Siena, Créteil, Torino, Padova, Milano, Messina, Newcastle

11.30 | Prenatal titinopathies are the severe end of the TTN-related myopathies spectrum: genotype- phenotype correlation on a large international cohort
M. F. Di Feo, A. Brady, F. Forzano, M. Iascone, P. D'Oria, L. Spaccini, S. Kurbatov, E. Giorgio, G. Casalis Cavalchini, A. Brusco, B. Udd, M. Savarese.
Helsinki, Middlesex, Londra, Bergamo, Milano, Voronezh, Pavia, Torino

11.45 | Whole Exome Sequencing of 126 patients highlights variants in candidate genes associated with FSHD
C. Strafella, D. Megalizzi, V. Caputo, G. Trastulli, L. Colantoni, S. Bortolani, E. Torchia, M. Monforte, C. Caltagirone, E. Ricci, G. Tasca, E. Giardina, R. Cascella.
Roma, Newcastle, Tirana

12.00 | When it is not FSHD: reviewing the mimics
G. Gadaleta, L. Vercelli, G. Urbano, E. Rolle, R. Tupler, T. Mongini.
Torino, Modena

12.15 | New insights for the interpretation of SMCHD1 gene variants effect: a functional study on a heterogeneous cohort of patients
S. Pini, M.Francesca Di Feo, M. Chiara, M.Savarese, P. Uva, M. Iascone, F. Santorelli, M. Vandroux, J. Laporte, A. Puliti, N. Albano, M. Seri, T. Pippucci, F. Isidori, A. Cereda, B. Udd, V. Salsi, R. Tupler.
Modena, Worcester, Genova, Milano, Bari, Helsinki, Bergamo, Pisa, Illkirch, Bologna, Bergamo, Vaasa

12.30 – 13.00 | MAIN LECTURE
Chaipersons: *Adele D'Amico (Roma), Tiziana Mongini (Torino)*

Gene Therapy for Rare Neuromuscular Disorders
Stefania Corti (Milano)

13.00 – 14.00 | Lunch

13:00 – 16:30 | POSTER VIEWING

14.00 - 15.00 | SYMPOSIUM NO CME

Myasthenia Gravis: Burden of disease and innovative treatments targeting the FcRn
Chairperons: *Lorenzo Maggi (Milano), Carmelo Rodolico (Messina)*

FcRn inhibition: a novel treatment option in myasthenia gravis and beyond
Raffaele Iorio (Roma)

Improvement in gMG health-related quality-of-life beyond the sole relief of signs and symptoms
Francesco Saccà (Napoli)

Conclusions
Lorenzo Maggi (Milano), Carmelo Rodolico (Messina)

15.00 – 16.00 | SYMPOSIUM NO CME

Get off the Miastenia Gravis rollercoaster through a continuos C5 inhibition
Chairpersons: *Giovanni Antonini (Roma), Rocco Liguori (Bologna)*

15:00 | Innovative therapies in gMG: the role of complement
Francesco Habetswallner (Napoli)

15:30 | A new gMG therapeutics algorithm in the precision medicine era
Renato Mantegazza (Milano)

16.00 - 16.30 | Coffee Break

16.30 - 18.30 | WORKSHOP
NOVEL THERAPEUTIC APPROACHES IN LIMB GIRDLE MUSCULAR DYSTROPHIES
Chairpersons: *Carlo Minetti (Genova), Vincenzo Nigro (Napoli)*

16.30 | Future development of gene therapy in sarcoglycanopathies and FKRP-related LGMD
Sophie Olivier (Paris, France)

17.00 | Novel therapeutic approaches in sarcoglycanopathies
Dorianna Sandonà (Padova)

17.30 | Gene Therapy for Sarcoglycanopathies
Amanda Haidet-Phillips (Ohio, USA)

18.00 | Therapies in LGMD2I/R9-FKRP related Limb Girdle Muscular Dystrophy
Douglas M. Sproule (North Carolina, USA)

18.30 - 19.30 | GENERAL ASSEMBLY

20:00 | SOCIAL DINNER

PETRARCA HALL

ORAL COMMUNICATIONS | METABOLIC MYOPATHIES
Chairpersons: *Giovanna Cenacchi (Bologna), Liliana Vercelli (Torino)*

11.00 | Exploring the role of Mobile Health Technologies in detecting subtle motor impairment in paucisymtomatic Late-onset Pompe Disease: a preliminary study
B. Labella, A. Rizzardi, S. Cotti Piccinelli, C. Zatti, C. Hansen, R. Romijnders, W. Maetzler, F. Caria, B. Risi, S. Damioli, E. Olivieri, L. Ferullo, L. Poli, A. Padovani, A. Pilotto, M. Filosto.
Brescia, Kiel

11.15 | Mitochondrial involvement in Patients with Autism spectrum disorders and Intellectual disability: a histological and genetic study
C. Scuderi, S. Santa Paola, M. Lo Giudice, M. Giuliano, F. D. Di Blasi, S. Città, R. Pettinato, G. A. Vitello, C. Romano, E. Borgione.
Troina

11.30 | Pregnancy in late-onset Pompe disease (LOPD) women: a multi-centre experience
O. Musumeci, L. Vercelli, M. Porcino, G. Gadaleta, I. G. Arena, T. Mongini, C. Rodolico, A. Toscano.
Messina, Torino

11.45 | Prevalence of Sleep Disorders in a cohort of patients with Primary Mitochondrial Diseases
I. G. Arena, C. Consulo, M. Porcino, C. Usbergo, C. Rodolico, A. Toscano, O. Musumeci.
Messina

12.00 | Clinico-pathologic features and molecular genetic spectrum in a cohort of adult patients with mtDNA maintenance disorders
M. Meneri, D. Piga, F. Magri, D. Velardo, P. Ciscato, S. Zanotti, M. Sciacco, G. P. Comi, D. Ronchi.
Milano

12.15 | Deriving harmonized follow-up in hyperCKemia using a digital web tool
B. Buchignani, F. Sansone, G. Marinella, G. Astrea, S. Frosini, D. Cassandrini, A. Tonacci, R. Conte, R. Battini, A. Rubegni, F. M. Santorelli and the InGene 2.0 group.
Pisa, Roma

PALLADIO HALL

ORAL COMMUNICATIONS | CHANNELOPATIES AND INFLAMMATORY MYOPATHIES
Chairpersons: *Giovanni Meola (Milano), Antonio Di Muzio (Chieti)*

11.00 | LAMOTRIGINE as an anti-myotonic agent in Myotonic Dystrophy typ 1: an open-label single center pilot study
S. Cotti Piccinelli, B. Risi, F. Caria, S. Damioli, E. Bertella, F. Garofali, N. Ait Allali, B. Labella, L. Poli, V. Bonito, A. Padovani, M. Filosto.
Brescia

11.15 | Therapeutic approaches to adult Ab anti-NXP2+ Dermatomyositis: a single center experience
G. Urbano, G. Gadaleta, F. Rumbolo, G. Mengozzi, S. Boschi, L. Chiadò-Piat, E. Rolle, L. Vercelli, T. Mongini.
Torino

11.30 | Refining the clinical spectrum and prognosis of granulomatous myositis from a large cohort of patients
A. Lauletta, L. de le Hoye, O. Benveniste, M. Garibaldi.
Roma, Brussels, Parigi

11.45 | Multiple inflammatory biomarkers assay can help distinguish between idiopathic inflammatory myopathies subtypes
M. Lucchini, V. De Arcangelis, M. Mirabella.
Roma

12.00 | Clinical and functional characterization of the novel Nav1.4 sodium channel mutation, p.T592I, found in a three-generation family
C. Campanale, P. Laghetti, I. Saltarella, C. Altamura, P. Bernasconi, R. Mantegazza, R. Brugnoni, P. Tacconi, J. F. Desaphy.
Bari, Milano, Cagliari

12.15 | Phenotypic variability of Andersen -Tawil Syndrome in a family sharing the same allelic *KCNJ2* gene mutation
M. E. Onore, E. Picillo, A. Torella, E. Cesarone, G. Piluso, V. Nigro, L. Politano.
Napoli

Cappella degli Scrovegni - Padova

MANTEGNA HALL

08.00 - 09.00 | MUSCLE CLUB
Chairpersons: *Serenella Servidei (Roma), Antonella Pini (Bologna)*

08.00 | Why am I cramping? Think beyond obvious
G. Tammam, E. Vegezzi, S. Gana, A. Pichiecchio, E. Alfonsi, G. Cosentino.
Pavia

08.12 | A rare case of adult-onset myopathy with facial involvement: expanding genotype-phenotype relationship and histopathological findings
S. Cotti Piccinelli, B. Risi, F. Caria, S. Damioli, E. Bertella, B. Labella, L. Poli, V. Bonito, G. Lanzi, A. Padovani, M. Filosto.
Brescia

08.24 | A case of myalgias and hyperCKemia after exercise
M. Villa, P. Riguzzi, G. Capece, V. Zangaro, E. Sogus, M. Penzo, D. Gorgoglione, D. Sabbatini, S. Vianello, L. Bello, E. Pegoraro.
Padova

08.36 | Diagnostic flowchart in an adult patient with asymmetric ptosis
D. Zoppi, R. Bencivenga, T. Fioretti, A. Russo, F. Masciarelli, R. Di Leo, M. Pezzella, R. Iodice, G. Esposito, F. Manganelli, L. Ruggiero.
Napoli

08.48 | A rare cause of congenital hypotonia: clinical aspects and molecular findings
A. Decio, R. Giorda, R. Cima, S. Marelli, E. Diella, D. Sacchi, C. Legnani, F. Villa, M.T. Bassi, M.G. D'Angelo.
Bosisio Parini

09.00 | MAIN LECTURE
Chairperson: *Stefano Previtali (Milano)*

Update about steroid and vamorolone theraphy in DMD
Michela Guglieri (Newcastle, UK)

09.30 – 10.00 | Coffee Break

10.00 – 11.30 | ORAL COMMUNICATIONS: DYSTROPHINOPATHIES
Chaiperson: *Fernanda Fortunato (Ferrara)*

10.00 | Dystrophin and utrophin quantitation by targeted mass spectrometry in Duchenne and Becker muscular dystrophy
P. Riguzzi, L. Bello, M. Penzo, M. Villa, G. Capece, V. Zangaro, D. Gorgoglione, S. Vianello, D. Sabbatini, E. Canessa, Y. Hathout, E. Pegoraro.
Padova, Binghamton

10.15 | Natural history of Becker muscular dystrophy: a retrospective multicentre study
D. Gorgoglione, D. Sabbatini, P. Riguzzi, A. Petrosino, M. Penzo, M. Villa, G. Capece, S. Vianello, G. S. Previtali, A. Zambon, C. Bruno, C. Panicucci, M. Traverso, A. Berardinelli, R. Nicotra, S. Parravicini, A. Gardani, A. Ferlini, F. Fortunato, E. M. Mercuri, M. Pane, C. Palermo, S. Servidei, C. Sancricca, M. Briganti, S. Messina, M. Sframeli, C. Allegra, A. D'Amico, M. Catteruccia, V. Sansone, A. Barp, M. G. D'Angelo, I. Moroni, F. Bruschi, A. Ardissone, R. Masson L. Maggi, A. Gallone, V. Nigro, E. Picillo, A. Pini, M. Giannotta, R. Battini, G. Marinella, T. Mongini, G. Gadaleta, G. Urbano, E. Rolle, F. Ricci, F. Magri, A. Lerario, G. Siciliano, G. Ricci, L. Bello, E. Pegoraro.
Padova, Pisa, Milano, Pavia, Genova, Ferrara, Roma, Messina, Trento, Bosisio Parini, Napoli, Bologna, Torino

10.30 | Robust preclinical data support development of DYNE-251 as a potential treatment for individuals with DMD mutations amenable to exon 51 skipping
E. Delage, E. Cotali, M. L. Naylor, C. A. Desjardins, R. Venkatesan, E. O'Donnell, J. Hall, R. Russo, S. Spring, K. Tang, J. W. Davis II, T. Weeden, S. Zanotti, C. Mix, B. Han, O. Beskrovnaya, A. Dugar.
Paris

10.45 | Definition of diaphragmatic sleep disordered breathing and clinical meaning in Duchenne Muscular Dystrophy
F. Trucco, M. Davies, A. A. Zambon, D. Ridout, F. Abel, F. Muntoni.
Londra, Milano

11.00 | Cardiomyopathy in Duchenne muscular dystrophy: progression of myocardial involvement and pharmacologic treatment in a large cohort of patients from a single tertiary referral Centre
M. Sframeli, L. Oreto, C. Stancanelli, C. Allegra, R. Materia, C. Zito, G. Di Bella, V. Tudisco, C. Rodolico, F. De Luca, S. Messina.
Messina

11.15 | RNA in situ hybridization reveals the spatial localization of DMD isoforms in human adult brain areas
M. S. Falzarano, M. Mietto, F. Fortunato, R. Selvatici, J. Morgan, R. Phadke, F. Catapano, F. Muntoni, A. Ferlini.
Ferrara, Londra

11.30 – 13.30 | ROUND TABLE

Meeting with Patient's Associations
Chairpersons: *Giacomo Pietro Comi (Milano), Federica Ricci (Torino)*

13.30 | Awards and conclusions
Giacomo Pietro Comi (Milano)

PETRARCA HALL

10.30 – 12.00 | ORAL COMMUNICATIONS | MYOPATHIES
Chairpersons: *Lucia Ruggiero (Napoli), Maurizio Moggio (Milano)*

10.00 | Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (Dista-Myo): baseline results
S. Bortolani, M. Di Bari, E. Torchia, E. Rolle, A. Vicino, M. Rabuffetti, A. Marzegan, M. Cheli, M. Monforte, E. Ricci, J. Y. Hogrel, S. Sacconi, T. E. Mongini, L. Maggi, G. Tasca.
Torino, Roma, Milano, Losanna, Parigi, Newcastle

10.15 | Gene editing in Emery-Dreifuss Muscular Dystrophy myoblasts
E. Cattin, E. Mattioli, E. Schena, F. Corradi, A. Pini, L. Maggi, C. Fiorillo, A. Recchia, G. Lattanzi.
Bologna, Milano, Genova

10.30 | Human mutated MYOT and CRYAB genes cause a myopathic phenotype in Zebrafish
G. Marchetto, E. Cannone, V. Guglielmi, C. Tobia, B. Gnutti, B. Cisterna, A. Barbon P. Tonin, M. Schiavone, G. Vattemi.
Verona, Brescia

10.45 | Myopathic Ehlers-Danlos Syndrome with Rectus femoris involvement: genetic heterogeneity behind COL12A1 dysfunction
M. Zanobio, A. Torella, P. Sabatelli, R. Zeuli, M. Scarpato, G. Piluso, L. Merlini, V. Nigro.
Bologna, Napoli

11.00 | Myofibrillar and distal myopathies: natural history of an Italian cohort of patients
M.Cheli, S. Bortolani, E. Rolle, A. Vicino, S. Bonanno, T. Enrica Mongini, G. Tasca, L. Maggi.
Milano, Torino, Roma, Newcastle

11.15 | Heterozygous truncating variants in DAG1 are associated with sporadic and familial isolated hyperCKemia
M. Traverso, S. Baratto, M. Di Duca, C. Panicucci, M. Iacomino, S. Casalini, M. Grandis, P. Striano, F. Zara, R. Barresi, C. Bruno, C. Fiorillo, M. Scala.
Genova, Venezia

PALLADIO HALL

10.00 -11.30 | ORAL COMMUNICATIONS| MYASTHENIA GRAVIS
Chairpersons: *Amelia Evoli (Roma), Matteo Garibaldi (Roma)*

10.00 | Efficacy of rozanolixizumab in generalised myasthenia gravis: subgroup analyses from the randomised Phase 3 MycarinG study
G. Antonini, T. Vu, A. Drużdż, J. Grosskreutz, A. A. Habib, R. Mantegazza, K. Utsugisawa, J. Vissing, R. Beau Lejdstrom, M. Boehnlein, T. Gasalla, F. Grimson, T. Tarancón, V. Bril.
Roma, Tampa, Poznań, Lübeck, Milano, Irvine, Hanamaki, Copenhagen, Monheim, Toronto, Madrid

10.15 | RAISE-XT: An interim analysis of safety and efficacy in an open-label extension study of zilucoplan in patients with myasthenia gravis
C. Antozzi, M. Freimer, M. Isabel Leite, A. Genge, Y. Hussain, R. Mantegazza, K. Utsugisawa, T. Vu, P. W. Duda, B. Boroojerdi, M. Vanderkelen, R. Lowcock, J. F. Howard Jr.
Milano, Columbus, Oxford, Montreal, Austin, Hanamaki, Tampa, Monheim, Chapel Hill, Braine-l'Alleud

10.30 | Long-Term Safety, Tolerability, and Efficacy of Subcutaneous Efgartigimod PH20 in Patients with Generalized Myasthenia Gravis: Interim Results of the ADAPT-SC+ Study
J. F. Howard Jr, G. Li, T. Vu, D. Korobko, M. Smilowski, K. Banaszkiewicz, L. Liu, S. Steeland, J. Noukens, B. Van Hoorick, J. Podhorna, Y. Li, K. Utsugisawa, F. Sacca, H. Wiendl, J. L. De Bleecker, R. Mantegazza, in collaboration with the ADAPT-SC Investigator Study Group.
Manchester, Chapel Hill, Tampa, Katowice, Novosibirsk, Kraków, Ghent, Napoli, Hanamaki, Münster, Milano

10.45 | In vitro effects of Rituximab on peripheral B cell subsets of patients with Myasthenia Gravis
J. Morroni, E. Sabatelli, S. Marini, S. Falso, R. Iorio.
Roma

11.00 | Lambert-Eaton myasthenic syndrome in a single centre cohort of patients: clinical, neurophysiological features and therapeutic management
A. Pugliese, A. Barbaccia, F. Biasini, O. Musumeci, S. Messina, A. Toscano, C. Rodolico.
Messina

11.15 | Early administration of Rituximab improves clinical outcome of Myasthenia Gravis: a single center cohort study
E. Latini, M. Maestri Tassoni, M. Guida, M.C. Caselli, G. Siciliano, R. Ricciardi.
Pisa

POSTER SESSION

ITALIAN ASSOCIATION
of MYOLOGY

Poster Session

SMA and motor neuron disorder

Chairpersons: *Megi Meneri (Milano), Federica Trucco (Milano)*

P1 - Newborn with SMA II treated with Zolgensma: between expectations and the real life after one year of follow-up
S. Siliquini, G. Salesi, E. Cesaroni, S. Cappanera, I. Cursio, G. Pantalone, C. Marini.
Ancona

P2 - Preliminary data on safety and efficacy of Risdiplam treatment in a small cohort of adult 5q spinal muscular atrophy
A. G. Nanni, G. Milella, G. Piccirilli, M. Ucci, S. Idrissi, A. Fraddosio, A. Introna, V. Scacco, M. Megna, I. Simone, E. D'Errico.
Bari

P3 - Profile of cognitive abilities in spinal muscular atrophy type II and III
B. Buchignani, G. Cicala, F. Moriconi, M. Ricci, A. Capasso, G. Coratti, J. Casiraghi, E. Albamonte, P. Cristofani, C. Cutrona, M. C. Pera, L. Antonaci, C. Roncoroni, D. Chieffo, V. A. Sansone, R. Battini, M. Pane, E. Mercuri.
Pisa, Roma, Milano

P4 - Nusinersen in Children with Spinal Muscular Atrophy: Experience of a Pediatric Pain Center
C. Agosto, A. Salerno, F. Benedetti, I. Maghini, A. Divisic, F. Rusalen, A. Zanin, A. Avagnina, G. Perilongo, F. Benini.
Padova

P5 - Motor unit number estimation in distal muscles of pediatric and adult patients with spinal muscular atrophy
V. Vacchiano, F. Morabito, C. Faini, R. Not, G. Scarpini, A. Pini, R. Liguori.
Bologna

P6 - Long-term comparative efficacy and safety of risdiplam versus nusinersen in children with Type 1 spinal muscular atrophy
N. Hawkins, G. Sajeev, R. Evans, A. Mahajan, D. A. Scott, J. Nam, E. Gaki, S. Sutherland, C. Kokaliaris
Oxford, Glasgow, Boston, London, Basel, Welwyn Garden City

P7 - Phenotypic spectrum of three unrelated SMA Italian patients with a compound heterozygosity for a deletion and rare missense mutation in SMN1 gene
A. Russo, D. Zoppi, T. Fioretti, R. P. Bencivenga, S. Vallone, V. Maiolo, R. Iodice, G. Esposito, L. Ruggiero.
Napoli

P8 - A novel mutation in *MYH14* associated with distal spinal muscular atrophy
P. Ajdinaj, M.G. Rispoli, L. Ferri, A. Tessa, F.M. Santorelli, A. Di Muzio.
Chieti, Pisa, Pescara

P9 - New challenges in the treatment era of Spinal muscular atrophy, a single centre experience with risdiplam
C. Dosi, E. Minacapilli, R. Zanin, M.T. Arnoldi , R. Masson.
Milano

P10 - The rehabilitation project and the motor functional scales in three patients with sma type 1 treated with nusinersen: an integrated approach to better understand new phenotypes
N. Sommella, R. Not, G. Scarpini, M. Giannotta, A. Pini, A. Cersosimo.
Bologna

P11 - Using icf-based functional profile to guide development of new measures for identifying treatment effects in sma patients receiving disease-modifying therapies
M.C. Oliva, C. Ferrante, I. Gallo, T. Vespino, A. Santoro, I. Maniglio, G. Barraco, F. Distante, A. Trabacca.
Brindisi

P12 - Preliminary results from application of EnduSMA protocol to detect endurance and fatigability in spinal muscular atrophy
R. Chiappini, G. Ricci, F. Torri, A. Govoni, L. Fontanelli, F. Magri, V. Vacchiano, M. Coccia, G. P. Comi, R. Liguori, G. Siciliano.
Ancona, Bologna, Milano, Pisa

P13 - A potential biomarker in Spinal Muscular Atrophy: serum creatinine in nusinersen treated patients SMA
G. Placidi, C. Dosi, R. Zanin, E. Ciusani, R. Masson
Milano

P14 - A case of SMA type 0: clinical findings and literature review
M. Capelli, G. Scarpini, V. Laganà, M. Giannotta, L. Landolina, J. Sarajlija, I. Donati, C. Graziano, G. Ancora, D. Cordelli, A. Pini.
Bologna, Cesena, Rimini

P15 - 3D-stem cell spinal cord model: a new tool to study risdiplam tool compound mechanisms and therapeutic effects for Spinal Muscular Atrophy
A. D' Angelo, P. Rinchetti, I. Faravelli, F. Beatrice, M. Nizzardo, F. Rizzo, L. Ottoboni, S. Corti.
Milano

P16 - SUNFISH Parts 1 and 2: 4-year efficacy and safety data of risdiplam in Types 2 and 3 SMA
M. Oskoui, J. W. Day, N. Deconinck, E. S. Mazzone, A. Nascimento, K. Saito, C. Vuillerot, G. Baranello, O. Boespflug-Tanguy, N. Goemans, J. Kirschner, A. Kostera-Pruszczyk, M. C. Pera, L. Servais, J. Braid, M. Gerber, K. Gorni, C. Martin, W. Y. Yeung, R. S. Scalco, E. Mercuri, on behalf of the SUNFISH Study Group.
Montreal, Palo Alto, Brussels, Ghent, Rome, Barcelona, Tokyo, Lyon, London, Milan, Paris, Leuven, Freiburg, Warsaw, Oxford, Basel, Welwyn Garden City

P17 - Risdiplam in type 2 and 3 Spinal Muscular Atrophy: results of a cohort of adult Italian patients
D. Zoppi, R. P. Bencivenga, S. Tozza, A. Russo, A. Nevano, R. Iodice, R. Dubbioso, G. Aceto, F. Manganelli, L. Ruggiero.
Napoli

P18 - Serum MyomiRs: potential biomarkers for adult SMA patients upon nusinersen treatment
C. Malacarne, F. Saraceno, R. Zanin, F. Andreetta, R. Mantegazza, L. Maggi, S. Marcuzzo, S. Bonanno.
Parma, Milano

P19 - Multidisciplinary pathway for the management of the first orally therapy for spinal muscular atrophy: a pediatric regional hospitals network experience
L. Pivato, C. Agosto, S. Paccagnella, F. Benedetti, F. Temporin, G. Perilongo, F. Venturini, F. Benini.
Padova

P20 - Clinical and demographic features of patients with SMA on treatment with risdiplam: the itaSMAc experience
E. Albamonte, L. Maggi, E. Pegoraro, S. Messina, M. Sframeli, A. Lizio, M. Pane, R. Masson, C. Dosi, A. D'Amico, A. Pini, G. Scarpini, F. Ricci, T. Mongini, A. Zanolini, G. Coratti, E. Mercuri, V. A. Sansone, on behalf of ItaSMAc.
Bologna, Messina, Milano, Padova, Roma, Torino

P21 - Pregnancy and therapy discontinuance in Spinal Muscle Atrophy: what we want to know. Discussion of a case report.
G. Ricci, L. Fontanelli, R. Chiappini, F. Torri, M. Gherardi, G. Siciliano.
Pisa

P22 - JEWELFISH: 24-month safety, pharmacodynamic and exploratory efficacy data in non-treatment-naive patients with SMA receiving treatment with risdiplam
C. Bruno, C. A. Chiriboga, T. Duong, D. Fischer, J. Kirschner, M. Scoto, E. Mercuri, M. Gerber, K. Gorni, H. Kletzl, I. Carruthers, C. Martin, T. Gidaro, F. Muntoni, on behalf of the JEWELFISH Study Group
Genova, Palo Alto, Basel, Freiburg, London, Rome, Welwyn Garden City

MYOPATHIES

Chairperson: *Andrea Barp (Trento)*

P23 - A novel frameshift mutation in *HSPB8* gene is responsible for a severe progressive myopathy with cardiac involvement
V. Tudisco, M. Sframeli, L. Bosco,V. Greco, R. Materia, C. Stancanelli, C. Allegra, M. Russo, I. Said, A. Lentini, S. Briuglia, F. Fattori, C. Rodolico, S. Messina.
Messina, Roma.

P24 - A novel mutation in *MCM3AP* gene expands the phenotype: a case report
M. Giannotta, F. Palombo, R. Pugliano, G. Scarpini, M.L. Valentino, R. Plasmati, F. Pastorelli, M. Maffei, D.M. Cordelli, A. Pini.
Bologna

P25 - *TMEM43* mutations: new variants and different phenotypes
D. Cassandrini, G. Marchetto, D. Lopergolo, A. Malandrini, C. Rodolico, P. Tonin, G. Vattemi, F.M. Santorelli.
Messina, Pisa, Siena, Verona

P26 - A novel mutation in LDB3 gene in a patient with Oculopharyngeal Muscular Dystrophy
G. Vadi, F. Torri, F. Baldinotti, G. Alì, G. Cenacchi, G. Ricci, G. Siciliano.
Bologna, Pisa

P27 - Clinical and molecular characterization of two siblings affected by Brody Myopathy
D. Velardo, S. Antognozzi, M. Rimoldi, S. Pagliarani, F. Cogiamanian, S. Barbieri, S. Corti, G. P. Comi, D. Ronchi.
Milano

P28 - Clinical and pathological findings in a couple of Italian Gypsy siblings with Charcot-Marie-Tooth type 4D and a review of the current literature
C.R. Ferrari Aggradi, S. Corti, D. Gagliardi, F. Magri, D. Velardo, G. P. Comi, E. Abati.
Milano

P29 - Centronuclear myopathy caused by a novel heterozygous *BIN1* variant: clinical, histological and MRI evaluation
F. Cortese, M. Brienza, L. De Giglio, A. Todisco, M. Garibaldi, F. Fattori, C. Bernardi, M.C. Altavista, E.M. Pennisi.
Roma

P30 - *HACD1*- Related myopathy with centronuclear myopathy and positive response to l-carnitine supplementation
G. Capece, P. Riguzzi, M. Villa, V. Zangaro, E. Sogus, M. Penzo, D. Gorgoglione, D. Sabbatini, S. Vianello, C. Bertolin, L. Bello, E. Pegoraro.
Padova

P31- Muscle MRI findings in a pediatric case of congenital fiber type disproportion associated with mutation in *TPM3* gene
S. Fusco, S. Parravicini, M. Paoletti, A. Iosca, M.I. Dainesi, C.A. Quaranta, R. Nicotra, V. Vacchini, A. Gardani, S. Gana, A. Berardinelli, A. Pichiecchio.
Pavia

P32 - X-linked Myopathy with Excessive Autophagy: Natural History of a French case series
G. Alfieri, G. Fernandez Eulate, I. Ackermann-Bonan, M. Spinazzi, F. Duval, G. Sole, A. Magot, F. Caillon, A. Pegat, E. Salort-Campana, A. Behin, T. Stojkovic.
Paris, Roma, Angers, Bordeux, Nantes, Lyon, Marseille

P33 - The Italian myotilinopathy population
A. Lupi, P. Riguzzi, S. Bortolani, M. Filosto, R. Liguori, S. Ravaglia, G. Ricci, F.M. Santorelli, P. Tonin, G.N. Vattemi, L. Maggi, G. Tasca, L. Bello, E. Pegoraro.
Bologna, Brescia, Crotone, Milano, Pavia, Padova, Pisa, Verona

P34 - Clinical and histological spectrum of axial myopathy: A retrospective study
G. U. Borin, F. Lanzafame, G. Marchetto, M. Tosi, S. Piffer, S. Gibertini, D. Cassandrini, F. Santorelli, L. Maggi, G. Vattemi, P. Tonin.
Milano, Verona, Pisa

P35 - Hyperckemia and CNS involvement in a 2 yrs old child: when the things get harder.
A. Iosca, S. Fusco, M.I. Dainesi, R. Nicotra, A. Gardani, S. Dogliani, S. Parravicini, V. De Giorgis, C. Varesio, S. Orcesi, A.L. Berardinelli.
Pavia

P36 - Primary triadopathies: the *STIM1*, *ORAI1* and *STAC3* players.
P. Riguzzi, L. Bello, A. Fusto, M. Villa, V. Zangaro, G. Capece, S. Tripodi, G. Minervini, M. Suman, S. Vianello, C. Agosto, G. Sorarù, C. Bertolin, V. Nigro, S. Tosatto, G. Cenacchi, R. Stramare, C. Reggiani, E. Pegoraro.
Bologna, Padova, Napoli

Myasthenia Gravis and Congenital Myasthenic Syndromes

Chaiperson: *Giovanni Antonini (Roma)*

P37- *ColQ*-Related Congenital Myasthenic Syndrome: A Clinical And Genetic Report
E. Olivieri, B. Risi, S. Cotti Piccinelli, F. Caria, S. Damioli, B. Labella, L. Ferullo, L. Poli, A. Padovani, M. Filosto.
Brescia

P38 - Long-Term Safety, Tolerability, and Efficacy of Efgartigimod in Patients with Generalized Myasthenia Gravis: Concluding Analyses from the ADAPT+ Study
M. Pasnoor, V. Bril, C. Karam, S. Peric, J. L. De Bleecker, H. Murai, A. Meisel, S. Beydoun, T. Vu, P. Ulrichts, B. Van Hoorick, C. T'joen, K. Utsugisawa, J. Verschuuren, R. Mantegazza, J. F. Howard Jr, in collaboration with the ADAPT Investigator Study Group.
Kansas, Toronto, Philadelphia, Belgrade, Ghent, Tokyo, Berlin, Los Angeles, Hanamaki, Leiden, Milan, Chape Hill

P39 - Usefulness and reliability of a self-monitoring diary for the management of patients with myasthenia gravis
F. Stragliati, A. Nuredini, P. Anceschi, S. Romano, S. Tinchelli, E. Chierici, E. Saccani.
Parma

P40 - Immune checkpoint inhibitors-induced Myasthenia Gravis: the need to stratify patients
E. Scarsi, S. Massucco, E. Faedo, C. Gemelli, M. Garnero, C. Genova, E. T. Tanda, C. Dellepiane, L. Benedetti, A. Schenone, M. Grandis.
Genova

P41 - Cancer frequency in muscle-specific tyrosine kinase (MuSK) myasthenia gravis
S. Falso, S. Marini, E. Sabatelli, A. Evoli, R. Iorio.
Roma

P42- Eculizumab for Immune Checkpoint Inhibitors (ICIs)-related Myasthenia Gravis
E. Rossini, A. Lauletta, P. Marchetti, G. Antonini, S. Morino, M. Garibaldi, L. Fionda.
Roma

P43 - Clinical and genetic heterogeneity in Congenital Myasthenic Syndromes: a pediatric case series
F. Trentin , B. Salce , G. Scarpini, M. Giannotta, G.M. Nocera, F. Pastorelli, F.M. Santorelli, F. Palombo, M.L. Valentino, D. Frattini, C. Fusco, V. Di Pisa, D.M. Cordelli, A. Pini.
Pisa, Bologna, Reggio Emilia

P44 - Lambert-Eaton myasthenic syndrome (LEMS) following SARS-CoV2 infection.
A. Barbaccia, A. Pugliese, F. Biasini, F. Andreetta, A. Migliorato, C. Rodolico.
Messina, Milano

P45 - Ravulizumab for the treatment of generalized myasthenia gravis: timing of response
R. E. Mantegazza , A. A. Habib, M. Benatar, T. Vu, A. Meisel, S. Attarian, M. Katsuno, S. Liao, K. N Beasley, J. F. Howard Jr.
Irvine, Miami, Tampa, Berlin, Marseille, Nagoya, Boston, Chapel Hill, Milan

P46 - Clinical characterization in juvenile myasthenia gravis: a single centre experience.
F. Bruschi, A. Ardissone, C. Ciano, C. Antozzi, R. Mantegazza, I. Moroni.
Milano

P47 - A triple seronegative, refractory patient with Generalized Myasthenia Gravis successfully managed with Efgartigimod
Y.M. Falzone, B. Sorrenti, C. Laurini, C. Strano, L. Bosco, P. Schito, M. Scarlato, R. Fazio, M. Filippi, S. Previtali.
Milano

P48 - Long-term safety and efficacy of Eculizumab in generalized Myasthenia gravis: the 7 years, experience of one patient at Sant'Andrea Hospital of Rome
A. Lauletta, L. Tufano, E. Rossini, S. Morino, M. Garibaldi, L. Fionda.
Roma

P49 - A novel mutation of ColQ in a case of Congenital Myasthenic Syndrome
A. Lupica, P. Alonge, A. Torrente, E. Borgione, C. Scuderi, G. Crescimanno, F. Brighina.
Palermo, Troina

P50 - Light sensitivity in myasthenia gravis: clinical characteristics, pupillometry findings and impact on quality of life
E. Sabatelli, L. Bonagura, S. Falso, S. Marini, G. Della Marca, R. Iorio.
Roma

P51 - Anti-FcRn treament with Efgartigimod in generalized Myasthenia Gravis
C. Antozzi, R. Frangiamore, E. Rinaldi, F. Vanoli, F. Andreetta, E. Ciusani, S. Bonanno, L. Maggi, A. Gallone, A. Pinna, R. Mantegazza.
Milano

Metabolic Myopathies

Chairperson: *Andrea Martinuzzi (Treviso)*

P52 - A NOVEL MISSENSE VARIANT IN THE ACADVL GENE IN A PATIENT WITH VLCAD DEFICIENCY: A CLINICAL AND GENETIC STUDY
F. Caria, S. Cotti Piccinelli, S. Damioli, B. Risi, E. Bertella, V. Bonito, B. Labella, L. Poli, A. Padovani, M. Filosto.
Brescia

P53 - Cardiopulmonary exercise testing as outcome measure in primary mitochondrial myopathy: comparison with other functional measures
I. G. Arena, C. Usbergo, C. Consulo, C. Rodolico, A. Toscano, O. Musumeci.
Messina

P54 - A novel compound homozygous *ACO2* mutation in an infant with fatal progressive encephalopathy: biochemical characterization on muscle tissue
A. Somà, F. S. Ricci, S. Stanga, M. Mezzanotte, C. Marinaccio, R. D'Alessandro, S. Sottemano, G. Morana, M. Spada, M. Boido, T. E. Mongini.
Torino

P55 - A longitudinal analysis in a cohort of juvenile patients with late onset Pompe disease (LOPD)
M. Porcini, O. Musumeci, I. G. Arena, C. Consulo, C. Usbergo, C. Rodolico, A. Toscano
Messina

P56 - The ER-MITO (Emilia Romagna-Mitochondrial) project: prevalence and genetics of Chronic Progressive External Ophthalmoplegia (CPEO) in an Italian region
M. L. Valentino, L. Caporali, C. La Morgia, F. Palombo, M. Romagnoli, C. Fonti, A. Maresca, R. Liguori, C. Zenesini , R. D'Alessandro, V. Carelli, on behalf of the ER-MITO study group.
Bologna

P57 - Accidental LOPD diagnosis in a young man with no clinical and instrumental evidence of neuromuscular dysfunction
M. Sciacco, S. Lucchiari, M. Garbellini, G. P. Comi, D. Ronchi.
Milano

P58 - Expanding the spectrum of clinical presentations associated with *COA8* pathogenic variants
S. Antognozzi, F. Magri, M. Meneri, M. Garbellini, S. Salani, F. Fortunato, M. Ripolone, S. Zanotti, P. Ciscato, M. Sciacco, V. Parente, S. Corti, G. P. Comi, D. Ronchi.
Milano

P59 - A primary cardiological phenotype caused by an inherited mtDNA single deletion: a case report from an Italian pedigree
P. Lopriore, C. Neuhofer, V. Montano, A. Meli, A. Lo Gerfo, M. A. Caligo, R. Berutti, R. Kopajtich, G. Siciliano, H. Prokisch, M. Mancuso.
Pisa, Munich, Neuherberg

P60 - Sudoscan in Pompe disease: a preliminary study
A. Lupica, P. Alonge, A. Torrente, V. Di Stefano, F. Brighina, G. Crescimanno
Palermo

LGMD and FSHD

Chairperson: *Giovanna Lattanzi (Bologna)*

P61 - Observational study: the quality of life in patients with alpha-sarcoglycan, beta-sarcoglycan and gamma-sarcoglycan gene mutation
B. Vola, M. Cerletti, Y. Torrente, C. Sanchez Riera, M. Bianchi.
London, Milano, Roma

P62 - A potential treatment for sarcoglycanopathy: pharmacological profile of C17 CFTR corrector and its efficacy with a new regimen of administration
A. Benetollo, M. Scano, S. Parrasia, L. Biasutto, F. Dalla Barba, M. Carotti, L. Nogara, B. Blaauw, D. Sandonà .
Padova

P63 - Validation of a blood-based assay for dysferlinopathy in a large Indian cohort
D. Cox, M. Henderson, Y. Parkhurst, P. Gaitonde, R. Dastur, S.S. Emmons, R. Barresi.
Newcastle, Seattle, Venice

P64 - A novel c.1478G>A calpain 3 gene variant associated to autosomal dominant calpainopathy
E. Faedo, S. Massucco, C. Gemelli, C. Fiorillo, M. Moggio, M. Ripolone, E. Bellone, S. Patrone, A. Mammi, A. Geroldi, A. Gaudio, P. Mandich, F. Gotta, E. Scarsi, A. Schenone, M. Grandis.
Genova, Milano

P65 - Facioscapulohumeral dystrophy: an epidemiologic investigation in Abruzzo
G. Polito, C. Ciprietti, S. Melchiorre, P. Adjnaj, A. Di Muzio.
Chieti

P66 - A four-years clinical follow-up and muscle MRI study in FSHD towards trials readiness
F. Torri, G. Aringhieri, L. Caldararo, S. Ortori, G. Siciliano, G. Ricci.
Pisa

P67 - A novel *LMNA* mutation causing atypical early-onset EDMD, with Achilles tendon contractures and limb-girdle weakness: diagnostic and therapeutic considerations
E. Minacapilli, C. Dosi, R. Zanin, F. Ricci, S. Gibertini, L. Maggi, R. Masson.
Milano, Torino

P68 - A novel case of *TRIM32*-related Limb Girdle Muscular Dystrophy
M. Rimoldi, F. Magri, S. Antognozzi, G. Romagnoli, D. Velardo, V. Parente, C. Cinnante, S. Corti, G. P. Comi, D. Ronchi.
Milano

P69 - A retrospective analisys of motor and respiratory progression in a population of *FSHD1* patients: how fast does the disease go?
G. Colacicco, E. Carraro, L. Martinelli, E. De Mattia, E. Roma, M. Croci, A. Lizio, V.A. Sansone.
Milano

P70 - *LGMDR19*: A MILD *GMPPB*-LINKED DISEASE PHENOTYPE
L. Ferullo, S. Cotti Piccinelli, F. Caria, B. Risi, S. Damioli, B. Labella, E. Olivieri, L. Poli, A. Padovani, M. Filosto.
Brescia

P71 - Calpain and Becker dystrophy: a double trouble
A. Lupica, P. Alonge, A. Torrente, E. Borgione, C. Scuderi, G. Crescimanno, F. Brighina.
Palermo, Troina

P72 - LMGD-D2 *TNPO3* related: A quality of life study
C. Angelini, A. A. Rodríguez.
Padova, Bilbao

P73 - Muscular and peripheral immunological profiles of patients affected by LGMDR3
C. Panicucci, A. Amaro, S. Pintus, F. Antonini, F. Reggiani, S. Baratto, E. Principi, C. Fiorillo, G. Del Zotto, D. Cassandrini, A. D'Amico, D. Lopergolo, A. Malandrini, E. Malfatti, G. Urbano, T. Mongini, A. Fusto, E. Pegoraro, S. Previtali, C. Rodolico, G. Tasca, U. Pfeffer, C. Bruno, L. Raffaghello.
Genova, Pisa, Roma, Siena, Créteil, Torino, Padova, Messina, Newcastle, Milano

Dystrophinopathies

Chairperson: *Emilio Albamonte (Milano)*

P74 - Feasibility of switch from prednisone to vamorolone in patients with DMD in VBP15-004 study
G. J. Van Daal, P. K. Clemens, E. C. Smith, I. Horrocks, R. Finkel, J. Mah, N. Deconinck, L. De Waele, V. Straub, G. Baranello, S. Spinty, A.M. Childs, J. Vilchez-Padilla, A. Nascimento-Osorio, E. Niks, I. de Groot, M. Katsalouli, E. Hoffman, M. Guglieri on behalf of the VISION-DMD.
Pratteln, Glasgow, Orlando, Calgary, Brussels, Newcastle, London, Leeds, Valencia, Barcelona, Nijmegen, Rockville, Leuven, Leiden

P75 - Efficacy and safety of vamorolone during 48-week treatment in patients with Duchenne Muscular Dystrophy (DMD) in VBP15-004 study
G. J. Van Daal, E. P. Hoffman, P.R. Clemens, S.J. Perlman, E.C. Smith, I. Horrocks, R.S. Finkel, J.K. Mah, N. Deconinck, N. Goemans, J. Haberlova, V. Straub, M.M. Ryan, M. Tulinius, R. Webster, H.J. McMillan, G. Baranello, S. Spinty, A.M. Childs, K.A. Selby, J.J. Vilchez-Padilla, A. Nascimento-Osorio, E.H. Niks, I.J.M. de Groot, M. Katsalouli, M. Guglieri on behalf of the VISION-DMD.
Pratteln, Pittsburgh, Durham, Orlando, Calgary, Brussels, Prague, Newcastle, Gothenburg, Ottawa, London, Liverpool, Vancuver, Valencia, Leiden, Athens, Sydney, Leuven, Barcelona, Nijmegen

P76 - Linkage analysis in a family with recurrence of duchenne muscular dystrophy defined the at risk haplotype and identified a clue of a more complex genomic rearrangement
M. Neri, L. Fiocco, F. Fortunato, M. Fabris, A. Margutti, C. Trabanelli, P. Rimessi, R. Selvatici, F. Gualandi, A. Ferlini.
Ferrara

P77 - Ergoreflex Sensitivity is Associated with Cardiac Involvement in Becker Muscular Dystrophy
V. Castiglione, P. Sciarrone, F. Torri, C. Borrelli, A. Barison, G. Todiere, C. Grigoratos, G. Siciliano, C. Passino, M. Emdin, G. Ricci, A. Giannoni.
Pisa

P78 - The spine fracture burden in boys with DMD treated with the novel dissociative steroid vamorolone versus deflazacort and prednisone
R. Rooman, M. Guglieri, S. Jackowski, U. Dang, M. Scharke, J. L. Jaremko, K. Koujok, M. Matzinger, N. Shenouda, K. Siminoski, M. Leinonen, S. Hasham, P. Clemens, M. McDermott, R. Griggs, E. Hoffman, L. Ward on behalf of the Ottawa Pediatric Bone Health Research Group, the FOR DMD Investigators of the Muscle Study Group and the CINRG VBP-//LTE Investigators.
Pratteln, Ottawa, New York, Newcastle, Rockville

P79 - Daily regimens of prednisone, deflazacort and vamorolone improve motor function similarly in patients with Duchenne Muscular Dystrophy
M. Guglieri, G.J. Van Daal, R. Rooman, C. McDonald, E. Henricson, A. Linden, P. Clemens, R. Griggs, P. Shieh, I. Horrocks, J. Mah, R. Finkel, N. Goemans, V. Straub, M. Ryan, H. McMillan, S. Spinty, E. Hoffman, on behalf of the VISION-DMD, FOR-DMD and CINRG investigators.
Newcastle, Pratteln, Sacramento, Pittsburgh, Los Angeles, Calgary, Memphis, Melbourne, Ottawa, Liverpool, Binghamton, New York, Leuven

P80 - Longitudinal forced vital capacity in Becker muscular dystrophy patients from the Padova cohort
M. Penzo, L. Bello, P. Riguzzi, M. Villa, G. Capece, D. Sabbatini, D. Gorgoglione, S. Vianello, A. Vianello, E. Pegoraro.
Padova

P81 - Retrospective study on the trend of NSAA and 6MWT scores of a cohort of DMD patients treated with cortisone according to age of initiation of therapy
M. I. Dainesi, A. Gardani, A. Iosca, S. Fusco, C.A. Quaranta, G. Catalano, R. Nicotra, L. Carraro, S. Parravicini, A. L. Berardinelli.
Pavia

P82 - DMD in females: a case series of eleven symptomatic carriers
G. D'Amario, I. Venezia, G. Cordaro, C. Guidetti, V. Napoli, A. L. Frongia, M. Pane, E. Mercuri.
Roma

P83 - Motor Functional findings and muscle MRI patterns of DMD patients with small mutations
C. Brogna, L. Cristiano, T. Verdolotti, G. Tasca, L. Ficociello, A. Frongia, N. Forcina, R. Ferrante, G. Stanca, M. Pane, E. Mercuri.
Roma

P84 - Genome-Wide Association Study for the identification of genetic modifiers of cardiomyopathy in Duchenne muscular dystrophy
D. Sabbatini, D. Gorgoglione, M. Penzo, S. Vianello, P. Riguzzi, M. Villa, A. Fusto, G. Capece, A. Berardinelli, C. Bruno, C. Panicucci, G. P. Comi, F. Magri, A. D'Amico, M. Catteruccia, L. Travaglioni, G. D'Angelo, V. Sansone, E. Albamonte, 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, M. Sframeli, G. Vita, R. Masson, S. Parravicini, G. Sorarù, L. Bello, E. Pegoraro.
Padova, Genova, Milano, Bosisio Parini, Torino, Roma, Napoli, Messina, Pavia

P85 - Poor Bone Health in Patients with Duchenne Muscular Dystrophy
E. Carraro, L. Martinelli, M. Pane, C. Palermo, A. Lizio, F. Trucco, E. Albamonte, E. Mercuri, V. A. Sansone.
Milano, Roma

Disease Mechanisms and Pathogenesis

Chairperson: *Linda Ottoboni (Milano)*

P86- Defining molecular and functional consequences of titin mutations in muscle progenitors from affected patients
C. Fiorillo, S. Baratto, S. Baldassari, E. Sondo, M. Traverso, G. Astrea, A. Biquand, E. Malfatti, M. Savarese, C. Bruno, N. Pedemonte.
Genova, Pisa, Parigi, Helsinki

P87 - Zebrafish models as new tools to study Sarcoglycanopathies
F. Dalla Barba, M. Carotti, A. Benetollo , M. Scano , P. Caccin, F. Argenton, D. Sandonà.
Padova

P88 - Circulating muscle-derived miR-206 links skeletal muscle dysfunction to heart sympathetic denervation
M. Ronfini, V. Di Mauro, V. Prando, A. Franco-Romero, A. Mazzaro, G. Favaro, F. Lo Verso, L. Dokshokova, A. Armani, G. Sorarù, M. Guescini, D. Catalucci, M. Mongillo, M. Sandri, T. Zaglia.
Padova, Milano, Urbino

P89 - Spastic paraparesis and myopathic signs associated to *POLRMT* mutations
F. Magri, S. Antognozzi, M. Meneri, F. Fortunato, M. Garbellini, M. Ripolone, A. Giacobbe, L. Napoli, G. P. Comi, D. Ronchi.
Milano

P90 - Whole miRNome profiling in myasthenia gravis thymus: novel insights into the molecular events underlying autoimmunity
M. C. Tarasco, N. Iacomino, L. Scandiffio, F. Bortone, S. Marcuzzo, S. Bonanno, R. Frangiamore, D. Pistillo, E. Voulaz, M. Alloisio, L. Maggi, C. Antozzi, R. Mantegazza, P. Cavalcante
Milano, Monza

P91 - MicroRNAs as biomarkers of responsiveness to immunosuppressive drugs in myasthenia gravis: towards personalized medicine
N. Iacomino, M. C. Tarasco, M. Ballardini, R. Frangiamore, F. Vanoli, L. Maggi, C. Antozzi, M. Foti, R. Mantegazza, P. Cavalcante.
Milano, Monza

P92 - Evaluation of aggrephagy markers in genetically defined myofibrillar myopathies, and patients
G. Riolo, E. Iannibelli, S. Gibertini, A. Ruggieri, A. Carnazzi, G. Tasca, S. Bortolani, L. Maggi.
Milano, Roma

P93 - Nuclear platform reorganization and nuclei orientation are lost in EDMD2 human myoblasts subjected to mechanical stimulation
V. Cenni, C. Evangelisti, S. Neri , P. Sabatelli, M. Cavallo, G. Lattanzi, E. Mattioli.
Bologna

P94 - Differential dysferlin expression in human and rat fast and slow skeletal muscle suggests different role
M. Meznaric, C. Angelini.
Padova

P95 - Transcriptomic and morphological alterations in hTNPO3 MUT-microinjected Zebrafish embryos modelling Limb Girdle Muscular Dystrophy D2 in vivo
M. Bergonzoni, M.T. Rodia, R. Costa, S. Pacilio, M. Fazzina, R. Casadei, F. Frabetti, G. Cenacchi.
Bologna, Rimini

P96 - Emerging role of myomiRs as biomarkers and therapeutic targets in Emery-Dreifuss muscular dystrophies
S. Bonanno, C. Malacarne, E. Mattioli, F. Saraceno, P. Cavalcante, L. Santovito Spiro, M. Cheli, C. Fiorillo, F. Andreetta, R. Mantegazza, S. Marcuzzo, L. Maggi, G. Lattanzi.
Bologna, Chicago, Genova, Parma

P97 - Microbiota Dysbiosis influences Immune System and Muscle Pathophysiology of Dystrophin Deficient Mice
A. Farini, L. Tripodi, C. Villa, F. Strati, B. Cassani, F. Caprioli, F. Facciotti, M. Quattrocelli, Y. Torrente.
Milano, Cincinnati

P98 - Efficient decellularization method for the accomplishment of engineered human skeletal muscle
S. D'Agostino, F. Magarotto, G. Tafuro, G. Germano, C. Biz, A. Semenzato, P. Gamba, M. Muraca, M. Pozzobon.
Padova

Diagnostic and Therapeutic tools

Chairperson: *Rita Barresi (Venezia)*

P99 - Attenuated total reflection-Fourier transform infrared spectroscopy (ATR-FTIR) in muscle diseases
G. Primiano, C. Sancricca, A. Primiano, J. Gervasoni, A. Urbani, F. Marini, A. Sabino, R. Calvani, S. Servidei.
Roma

P100 - Eye tracking communication device opens world of communication for a non-verbal child affected by SMARD 1
A. Ferrero, N. Gironella, F. Re, A. Ubaldi, M. Coccia.
Ancona

P101 - Sympathetic neuropathology is revealed in muscles affected by Amyotrophic Lateral Sclerosis
A. Mazzaro, V. Vita, M. Ronfini, I. Casola, A. Klein, G. Dobrowolny, G. Sorarù, A. Musarò, M. Mongillo, T. Zaglia.
Padova, Roma

P102 - A new 6MWT web-tool to facilitate clinical monitoring of neuromuscular patients
G. Marinella, B. Buchignani, A. Rubegni, S. Frosini, E. Dati, A. Tonacci, R. Conte, F. Sansone, G. Diodato, M.C. Scudellari, F. Torri, G. Ricci, G. Siciliano, R. Battini, G. Astrea, F. M. Santorelli.
Pisa, Roma

P103 - Proposal for a model of care in a pediatric neuromuscular clinic: experience in Trento area
V. Fiorito, A. Iodice, G. Zappa, U. Pradal, R. Zuccarino.
Trento

P104 - Challenging diagnosis between an atypical phenotype of motor neuron disease and myositis: a case report.
L. Becattini, G. Vadi, F. Bianchi, L. Fontanelli, B. Giovannini, C. Meoni, G. Ricci, G. Siciliano.
Pisa

P105 - High plasma p-tau181 levels in amyotrophic lateral sclerosis: clinical-electrophysiological correlations and longitudinal trajectories
V. Vacchiano, A. Mastrangelo, C. Zenesini, S. Baiardi, P. Avoni, B. Polischi, S. Capellari, F. Salvi, R. Liguori, P. Parchi.
Bologna

P106 - An exploratory study on the use of a high-content image analysis system in myopathies
F. Torri, C. Filipponi, B. Ciurli, M. Lai, G. Alì, G. Vadi, M. Pistello, G. Siciliano, G. Ricci.
Pisa

P107 - Exploratory study of a passive wearable device (Exoband) as walking aid in neuromuscular patients
C. Semplicini, M. Agostini, C. Andrigo, V. Notararigo, S. Masiero, F. Piccione, G. Sorarù.
Padova

P108 - Diaphragmatic ultrasound: a promising technique for respiratory assessment of patients with facio-scapulo-humeral muscular dystrophy (FSHD)
E. Torchia, S. Bortolani, R. Inchingolo, A. Smargiassi, M. Monforte, G. Tasca, M. Bonini, L. Richeldi, E. Ricci.
Roma

P109 - Different approaches to the genetic diagnosis of hereditary peripheral neuropathies: a 2 years multicenter study
L. Bosco, M. Catteruccia, D. Diodato, M. Tosi, E. Pennisi, M. Garibaldi, S. Pro, E. S. Bertini, A. D'Amico, F. Fattori.
Roma

P110 - Reduction of Body Cellular Mass and Body Cellular Mass Index as an early marker of sarcopenia in Amyotrophic Lateral Sclerosis patients
G. Greco, L. Boffa, M. Goglia, S. Seraceno, A. M. Ruggieri, G. Merra, L. Romano, E. Frezza, G. Vietri, F. Gruosso, G. Nardino, I. Petitta, P. Leuratti, A. De Lorenzo, R. Massa.
Roma

P111 - Unravelling combined RNA interference and gene therapy in vitro and in vivo disease models as a potential therapeutic strategy for CMT2A
F. Rizzo, S. Bono, M.D. Ruepp, S. Salani, L. Ottoboni, E. Abati, V. Melzi, C. Cordiglieri, S. Pagliarani, R. De Gioia, A. Anastasia, M. Taiana, M. Garbellini, S. Lodato, P. Kunderfranco, D. Cazzato, D. Cartelli, C. Lonati, N. Bresolin, G. P. Comi, M. Nizzardo, S. Corti.
Milan, London

P112 - D4Z4 methylation analysis combined with machine learning pipelines: a novel tool for identifying FSHD subjects
V. Caputo, C. Strafella, G. Trastulli, D. Megalizzi, C. Fabrizio, A. Termine, L. Colantoni, J. Gimenez, M. Monforte, C. Caltagirone, E. Ricci, G. Tasca, R. Cascella, E. Giardina.
Rome, Newcastle

P113 - MethylSeq based assay to assess the epigenetic setting of D4Z4 repetitive elements in facioscapulohumeral muscular dystrophy
V. Salsi, M. Chiara, S. Pini, L. Ruggiero, F. Santorelli, S. C. Previtali, M. G. D'Angelo, C. Rodolico, S. Bonanno, L. Maggi, R. Tupler.
Modena, Worcester, Bari, Napoli, Pisa, Milano, Messina, Bosisio Parini

P114 - The case of Friedreich Ataxia as an example to exploit the pleiotropic effects of nanoparticles
C. Villa, M. Mistretta, A. Farini, Y. Torrente.
Milano

P115 - Pediatric Palliative Care is the answer for met and unmet needs of children with neuromuscular diseases
C. Agosto, F. Benedetti, I. Maghini, P. Lazzarin, F. Benini.
Padova

P116 - IN VITRO ELECTROSTIMULATION ASSAY FOR FUNCTIONAL EVALUATION OF ENGINEERED SKELETAL MUSCLE TISSUE
S. Pacilio, T. Cramer, R. Costa, V. Papa, M. T. Rodia, B. Fraboni, M. L. Focarete, G. Cenacchi.
Bologna

P117 - Unraveling the role of high-resolution ultrasonography of peripheral nerves in adult spinal muscular atrophy
S. Bonanno, G. Devigili, R. Togni, M. Corradi, P. Lanteri, R. Mantegazza, R. Eleopra, L. Maggi.
Milano

P118 - Health360 Trials: a software module to manage clinical trials and patients from different clinical institutions
F. Sansone, G. Diodato, M. C. Scudellari, F. M. Santorelli, G. Siciliano, A. Malandrini, S. Matà, C. Angelini, A. Tonacci, The InGene. Project Group, R. Conte.
Pisa, Siena, Firenze, Roma, Padova

Congenital Myopathies

Chairperson: *Caterina Agosto (Venezia)*

P119 - Novel *PIEZO2* variants are responsible for a very suggestive clinical phenotype in early childhood
D. Diodato, M. Catteruccia, A. Sancesario, M. Tosi, L. Bosco, F. Fattori, G. De Luca, I. Mizzoni, A. Carlesi, E. Bertini, A. D'Amico.
Roma

P120 - Cognitive, adaptive and perseverative aspects characterization of children with XLMTM: an explorative study
F. Cumbo, M. Tosi, A. Carlesi, I. Mizzoni, M. Catteruccia, E. S. Bertini, A. D'Amico.
Roma

P121 - Collagen VI-related myopathies with onset in pediatric age: the value of clinical findings when not all the diagnostic pieces fit in
G. Guardi, M. Giannotta, G. Scarpini, M. L. Valentino, M. Maffei, F. Palombo, A. Ferlini, D.M. Cordelli, F. Gualandi, A. Pini.
Bologna, Ferrara

P122 - Congenital myopathy as a phenotypic expression of *CACNA1S* gene mutation: Case Report and literature systematic review
G. Marinella, A. Orsini, M. Scacciati, E. Costa, G. Astrea, S. Frosini, R. Pasquariello, A. Rubegni, G. Sgherri, M. Corsi, A. Bonuccelli, D. Peroni, R. Battini.
Pisa

P123 - Causative variants profile in a large Italian cohort of Collagen VI , related dystrophy
L. Fiocco, M. Neri, A. Margutti, F. Fortunato, A. D'Amico, A. L. Berardinelli, A. Pini, C. Fusco, C. Rodolico, C. Fiorillo, C. Bruno, C. Cereda, E. Pegoraro, E. Bertini, E. Ricci, E. Picillo, E. Mercuri, F. Vercellino, F. Massaro, G. Ciana, I. Moroni, L. Boccone, L. Maggi, L. Morandi, L. Merlini, L. Politano, M. Scutifero, M. Pane, M. Pedemonte, M. Filosto, M. Del Mastio, M. Traverso, P. Gasparini, P. Boffi, R. Massa, S. Messina, T. E. Mongini, R. Selvatici, A. Ferlini, F. Gualandi.
Ferrara, Roma, Pavia, Reggio Emilia, Messina, Genova, Padova, Napoli, Prato, Udine, Milano, Cagliari, Bologna, Firenze, Trieste, Torino

P124 - Novel *RYR1* pathological variants discovered in italian patients with dusty core disease
D. Piga, S. Zanotti, F. Magri, S. Salani, L. Napoli, M. Ripolone, F. Fortunato, D. Cassandrini, F. Fattori, M. G. D'Angelo, E. Albamonte, V. Nigro, S. Corti, G. P. Comi.
Milano, Pisa, Roma, Lecco, Napoli

Channelopathies and Inflammatory Myopathies

Chairperson: *Michela Coccia (Ancona)*

P125 - High dose intravenous corticosteroids in refractory Immune Checkpoint Inhibitor-related Myocarditis/Myositis/Myasthenia overlap syndrome
L. Tufano, A. Lauletta, S. Morino, B. Musumeci, G. Antonini, M. Garibaldi.
Roma

P126 - Cognitive function assessment in a cohort of patients with Myotonic Dystrophy Type 1: a single-center experience
B. Risi, S. Cotti Piccinelli, E. Ferrari, F. Caria, S. Damioli, E. Bertella, B. Labella, L. Poli, V. Bonito, A. Padovani, M. Filosto.
Brescia

P128 - Myotonic Dystrophy type 1: motor and cardio-respiratory evolution trajectories
E. Diella, A. LoMauro, M. Delle Fave, G. Canella, G. Meola, R. Cima, M. G. D'Angelo.
Milano, Bosisio Parini

P129 - A novel hypokalemic periodic paralysis mutation in the *SCN4A* gene shared by two families and description of a homozygous patient
S. Pagliarani, V. Sansone, F. Gerardi, M. Scarlato, S. C. Previtali, S. Lucchiari, G. Meola, G. P. Comi.
Milano, Bergamo

P130 - Beyond diagnostic criteria: the pitfalls in the diagnosis of inflammatory myopathies, a case series
L. Bosco, Y.M. Falzone, V.Canti, P. Rovere Querini, U. Del Carro, M. Filippi, S.C. Previtali.
Milano

P131 - Compound *CACNA1S* heterozygosity resulting in a novel phenotype of congenital myopathy and early onset periodic paralysis: report of two probands
S. K. Aburahma, M. Shboul, L. A. Rousan, S. Lucchiari, F. Biella, G. P. Comi, G. Meola, S. Pagliarani.
Irbid, Milano

P132 - A case of myasthenia and myositis after Covid vaccine
G. Vadi, F. Torri, M. Maestri, G. Alì, M. Lencioni, G. Siciliano, G. Ricci.
Pisa

P133 - Novel *HSPG2* gene mutation causing Schwartz Jampel syndrome in a Moroccan family.
D. Marelli, R. Brugnoni, C. Cappelletti, N. Iacomino, E. Canioni, L. Maggi, A. Ardissone.
Milano

P134 - A case of Amyotrophic Lateral Sclerosis initially misdiagnosed for myositis
C. Meoni, F. Bianchi, L. Becattini, L. Fontanelli, B. Giovannini, G. Siciliano.
Pisa

P135 - Long-term treatment and follow up in adult patients with anti-HMGRC immune-mediated myopathy
L. Vercelli, G. Gadaleta, G. Urbano, G. Brodini, L. Chiadò-Piat, F. Rumbolo, T. Manetta, T. Mongini.
Torino

P136 - Pre- and post-natal outcomes in congenital and childhood onset DM1 - the impact of parental diagnostic delay
F. Trucco, A. di Bari, F. Salmin, A. Lizio, E. Albamonte, A. Zanolini, M. Beretta, J. Casiraghi, L. Antonaci, R. de Sanctis, A. Salvalaggio, M. Catteruccia, M. Tosi, G. Marinella, R. Danti, F. Bruschi, A. Ferrero, B. Risi, A. Barp, M. Veneruso, C. Fiorillo, A. Berardinelli, A. Pini, R. Zuccarino, M. Filosto, M. Coccia, I. Moroni, G. Astrea, A. D'Amico, F. Ricci, M. Pane, E. Mercuri, V. Sansone.
Milano, Torino, Roma, Pisa, Ancona, Brescia, Trento, Genova, Pavia, Bologna

P138 - Myopathy in the oncologic patient: move beyond muscle inflammation
R. Costa, V. Di Pisa, R. D'Angelo, S. Rossi, R. Rinaldi, G. Cenacchi.
Bologna

P139 - Inclusion body myositis functional rating scale in IBM patients: a cross-sectional study
A. Vicino, M. Cheli, S. Bonanno, L. Maggi.
Milano, Lausanne

P140 - Preclinical Data Support the Initiation of the ACHIEVE Trial of DYNE-101 in Individuals with Myotonic Dystrophy Type 1 (DM1)
E. Delage, E. Cotali, D. Wolf, T. Picariello, L. Schlaefke, R. Russo, A. Chang, S. Hildebrand, J. Najim, Q. Qiu, T. Weeden, J. W. Davis II, C. Mix, B. Han, S. Zanotti, O. Beskrovnaya, W. Farwell.
Paris, Waltham

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- Thursday, 8th June from 09.00 to the end of the works
- Friday, 9th June from 07.15 to the end of the works
- Saturday, 10th June from 07.30 to the end of the works

AIM SECRETARIAT

The AIM Secretariat Desk will be present at the congress venue (next to the registration area) and will follow the following times

- June 8th from 10:00 to the end of the works
- June 9th from 08:30 to the end of the works

REGISTRATION

The meeting is reserved for a maximum number of 450 participants.
Participation in the event is subject to online registration by Monday 29 May 2023.
After the deadline, it will be possible to register directly at the congress venue.

	By 8th May 2023	From 9th May 2023
AIM MEMBER	**234,00 €**	**275,00 €**
AIM MEMBERS UNDER 35	**152,00 €**	**193,00 €**
NOT AIM MEMBER	**328,00 €**	**394,00 €**

VAT (22%) excluded

XXIII NATIONAL CONGRESS
OF ITALIAN ASSOCIATION of MYOLOGY

The registration fee includes:
Admission to scientific sessions
Congress Kit
Lunches and coffee breaks
Welcome Cocktail | 8th June 2023
Social Dinner | 9th June 2023
Certificate of attendance
CME certificate (it will be sent after the conclusion of the event after verifying the attendance and learning requirements)

PAYMENT METHOD
Online - Bank transfer and credit card
Onsite - ATM and credit card

CANCELLATIONS AND REFUNDS
Cancellations received by 21 April 2023 entitle to a refund of the amount paid, less 30% for administrative costs.
Cancellations received after this date will not give the right to any refund.
All refunds will in any case be made after the Congress has taken place.

BADGE
At the time of registration, each registered participant will be given a name badge which must always be shown in order to access the classrooms where the scientific sessions are held, the exhibition area, the catering services and the Welcome Cocktail on Wednesday 8th June.

CERTIFICATE OF ATTENDANCE
The certificate of participation can be downloaded online after completing the evaluation questionnaire. The Organizing Secretariat will send an e-mail to all participants on 10th June containing useful information to download it.

CLOAKROOM AND LUGGAGE STORAGE
A cloakroom and left-luggage office will be made available.

WELCOME COKTAIL
The Welcome Cocktail will be held on Wednesday 8 June at 20:00 on the third floor of the Padova Congress. Please wear your personal Congress badge to access the restricted area.

SOCIAL DINNER
The social dinner will be held on Friday 9th June at 20:45 at:

VILLA MOLIN
Via Ponte della Cagna 106 - Padova

Entry will be permitted only upon presentation of the relative invitation received at the time of registration for the Congress.

Accreditamento ECM

Il **XXIII Congresso Nazionale Associazione Italiana di Miologia**, Padova 8-10 giugno 2023 è inserito nel Piano Formativo anno 2023 First Class - Provider nr. 362 ed è stato accreditato presso la Commissione Nazionale per la Formazione Continua con **Obiettivo Formativo:**

18 - Contenuti tecnico-professionali (conoscenze e competenze) specifici di ciascuna professione, di ciascuna specializzazione e di ciascuna attività ultra-specialistica, ivi incluse le malattie rare e la medicina di genere per le seguenti figure professionali:

PROFESSIONE	DISCIPLINE
Farmacista	*Farmacia Ospedaliera; Farmacia Territoriale;*
Biologo	*Biologo*
Logopedista	*Logopedista*
Terapista Occupazionale	*Terapista Occupazionale*
Psicologo	*Psicoterapia; Psicologia;*
Medico Chirurgo	*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*
Infermiere Pediatrico	*Infermiere Pediatrico*
Fisioterapista	*Fisioterapista*
Infermiere	*Infermiere*
Dietista	*Dietista*
Tecnico di Neurofisiopatologia	*Tecnico di Neurofisiopatologia*
Tecnico Sanitario di Laboratorio Biomedico	*Tecnico Sanitario di Laboratorio Biomedico*
Terapista della Neuro e Psicomotricità dell'età Evolutiva	*Terapista della Neuro e Psicomotricità dell'età Evolutiva*

Il congresso è stato accreditato per un numero massimo di n. 450 partecipanti. Oltre tale numero e per professioni/ discipline differenti da quelle accreditate non sarà possibile rilasciare i crediti formativi. Si precisa che i crediti verranno erogati a fronte di una partecipazione del 90% ai lavori scientifici (farà fede esclusivamente l'orario di ingresso e uscita riportato nel foglio firme) e del superamento della prova di apprendimento con almeno il 75% delle risposte corrette.

L'evento, ID ECM 362-383225, ha ottenuto 4,8 crediti formativi.

Exhibition area

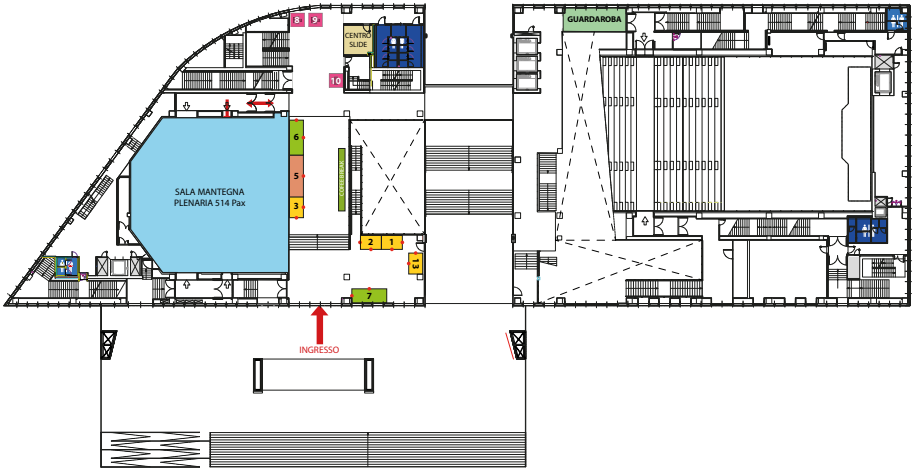
Padova Congress

GROUND FLOOR

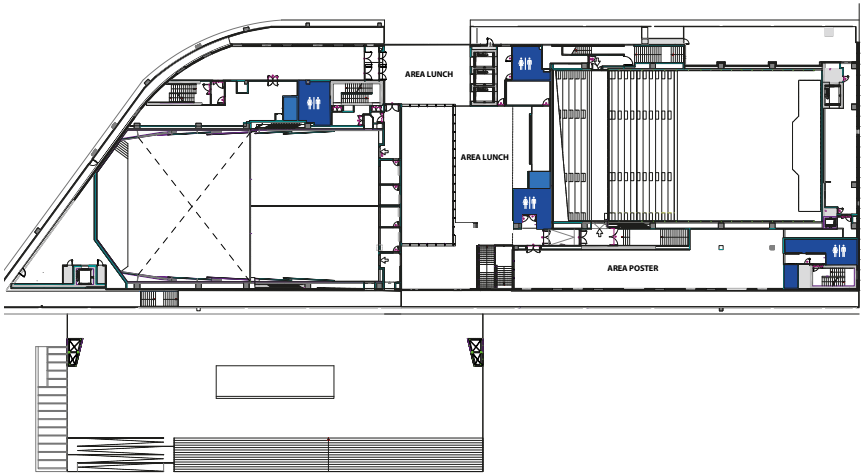
Exhibition area

Padova Congress

FIRST FLOOR



THIRD FLOOR



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ASSOCIAZIONE ITALIANA
MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

XXIII NATIONAL CONGRESS
OF ITALIAN ASSOCIATION of MYOLOGY

PADOVA
8th - 10th June 2023

www.congressoaim2023.it

ORGANIZING SECRETARIAT

FIRST CLASS
EVENTS & CONFERENCES

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