

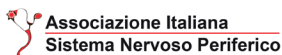


AIM 2025
TAORMINA

XXV NATIONAL CONGRESS
OF ITALIAN ASSOCIATION
OF MYOLOGY

Palazzo dei Congressi di Taormina
28 - 31 Maggio 2025

Il XXV National Congress of Italian Association of Myology
si svolge con i patrocini di:





INDICE

- pag. 01** | Consiglio Direttivo
- 02** | Faculty
- 35** | Poster Session

PROGRAMMA

- pag. 05** | Wednesday, May 28
- 11** | Thursday, May 29
- 20** | Friday, May 30
- 30** | Saturday, May 31

PROGRAM AT A GLANCE

Wednesday, May 28, 2025

	Plenary Room (floor 0)	Parallel Room 1 (floor -1)	Parallel Room 2 (floor 4)
13:00	REGISTRATIONS OPENING		
14:30 - 15:00	Greetings and Introduction 25 years of AIM		
15:00 - 15:30	Main Lecture Advancements in Therapeutics in Muscular Dystrophies		
15:30 - 17:00	Plenary Workshop 1 Exercise and training in muscular dystrophies		
17:00 - 17:30	Coffee Break		
17:30 - 18:30	ORAL COMMUNICATION DUCHENNE MUSCULAR DYSTROPHY (DMD)	ORAL COMMUNICATION MYASTHENIA GRAVIS	ORAL COMMUNICATION METABOLIC MYOPATHIES
18:30 - 19:15	Symposium Sponsored by SANTHERA NO CME		
19:15 - 19:45	Lecture sponsored by NOVARTIS NO CME		

Thursday, May 29, 2025

08:00 - 08:30	Lecture 1 Immunotherapy in MG from Multi-Target to Target-Specific Therapies		
08:30 - 10:00	Parallel Workshop 1 Update on myositis	Parallel Workshop 2 Congenital myopathies	
10:00 - 10:30	Lecture 2 Phenotypes and treatments of TK2 deficiency		
10:30 - 11:00	Coffee Break		
11:00 - 12:30	Plenary Workshop 2 Controversies in Mitochondrial Diseases: Debates and Emerging Perspectives		
12:30 - 13:30	Lunch and GDS Meetings		
13:30 - 14:15	NO CME PROGRAM	Symposium Sponsored by ROCHE NO CME	
14:15 - 15:00		Symposium Sponsored by BIOGEN NO CME	
15:00 - 15:45		Symposium Sponsored by DYNE THERAPEUTICS NO CME	
15:45 - 16:15	Coffee Break		
16:15 - 17:30	ORAL COMMUNICATION MYASTHENIA GRAVIS	ORAL COMMUNICATION MUSCULAR DYSTROPHIES	ORAL COMMUNICATION EXPERIMENTAL THERAPIES, MITOCHONDRIAL DISORDERS, CONGENITAL MYOPATHIES
17:30 - 19:00	Parallel Workshop 3 Pregnancy and childbirth with neuromuscular diseases	Parallel Workshop 4 FSHD: Approaches to Clinical Diagnosis and Phenotyping	

Friday, May 30, 2025

	Plenary Room (floor 0)	Parallel Room 1 (floor -1)	Parallel Room 2 (floor 4)
08:30 - 09:00	NO CME PROGRAM	Breakfast Seminar Sponsored by ITALFARMACO NO CME	
09:00 - 10:30	Parallel Workshop 5 MRI in muscular disorders	Parallel Workshop 6 Emergencies in Neuromuscular Disorders	
10:30 - 11:00	Coffee Break		
11:00 - 12:30	Parallel Workshop 7 New therapeutic roads in muscle disorders	Parallel Workshop 8 The Role of Calcium in Neuromuscular Disorders	
12:30 - 13:30	Lunch and GDS Meetings		
13:30 - 14:15	NO CME PROGRAM	Symposium Sponsored by UCB NO CME	
14:15 - 15:00		Symposium Sponsored by ARGENX NO CME	
15:00 - 15:45		Symposium Sponsored by ALEXION NO CME	
15:45 - 17:30	Coffee Break with Poster Session NO CME		
16:30 - 17:30	MUSCLE TRIVIAIM CONTEST Qualification		
17:30 - 18:30	ORAL COMMUNICATION GENETICS	ORAL COMMUNICATION SPINAL MUSCLE ATROPHY (SMA)	ORAL COMMUNICATION FSHD
18:30 - 20:00	General Assembly		

Saturday, May 31, 2025

08:00 - 09:00	GDS Meetings		
09:00 - 09:30	Lecture 3 In vitro platforms to study neuromuscular diseases: challenges and opportunities	Lecture 4 Updates on clinical trials in Mitochondrial Myopathies	
09:30 - 10:30	ORAL COMMUNICATION SPINAL MUSCLE ATROPHY (SMA)	ORAL COMMUNICATION MYOTONIC DYSTOPHIES	ORAL COMMUNICATION MYOSITIS AND OTHERS
10:30 - 11:00	Coffee Break		
11:00 - 12:00	MUSCLE TRIVIAIM CONTEST FINAL		
12:00 - 13:30	ROUND TABLE: MEETING with PATIENT's ASSOCIATIONS		
13:30 - 14:00	Closing ceremony & Awards		

**CONSIGLIO
DIRETTIVO**

**COMITATO
SCIENTIFICO**

**COMITATO
ORGANIZZATORE
LOCALE**



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Matteo Garibaldi, *Roma*
Marina Grandis, *Genova*
Francesco Habetswallner, *Napoli*
Maurizio Inghilleri, *Roma*
Giovanni Iolascon, *Napoli*
Raffaele Iorio, *Roma*
Amel Karaa, *Parigi*
Thomas Klopstock, *Monaco*
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Maurizio Gualtieri Moggio, <i>Milano</i>	Filippo Santorelli, <i>Pisa</i>
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Marcello Romano, <i>Palermo</i>	Ester Zito, <i>Milano</i>
Dario Ronchi, <i>Milano</i>	
Lucia Ruggiero, <i>Napoli</i>	
Elena Saccani, <i>Parma</i>	
Cristina Sancricca, <i>Roma</i>	

XXV AIM National Congress

PROGRAMMA

PLENARY ROOM

13:00 Registration

14:30 - 15:00 **Greetings and Introduction - 25 years of AIM**

AIM President

AIM Past Presidents

15:00 - 15:30 **Main Lecture**

Chairperson: *Antonio Toscano (Messina)*

Advancements in Therapeutics in Muscular Dystrophies

Giacomo Pietro Comi (Milano)

Plenary Workshop 1

Chairperson: *Giovanni Iolascon (Napoli)*

Exercise and training in muscular dystrophies

15:30 - 15:50 **Physiatric Approach**

Eric Voorn (Amsterdam)

15:50 - 16:10 **Pathophysiological Approach**

Mads Stemmerik (Copenhagen)

16:10 - 16:30 **Personalizing Exercise: Clinical Guidelines and Patient-Centered Outcomes in Muscular Dystrophies**

Gabriele Siciliano (Pisa)

16:30 - 17:00 **Discussion and Q&A**

17:00 - 17:30 **Coffee Break**

17:30 - 18:30 **ORAL COMMUNICATION | DUCHENNE MUSCULAR DYSTROPHY (DMD)**

Chairpersons: *Angela Lucia Berardinelli (Pavia), Andrea Barp (Trento)*

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42 **BONE HEALTH INDEX BY HAND X-RAY AS A TOOL TO MEASURE BONE FRAGILITY IN DMD**

Panicucci C.¹, Repetto A.¹, Angelelli A.², Casalini S.¹, Brolatti N.¹, Trucco F.³, Pedemonte M.³, Damasio M.B.⁴, Maghnie M.², Di Iorgi N.², Bruno C.¹

¹ Centre of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova, Italy; ²Department of Pediatrics, IRCCS Istituto Giannina Gaslini, Genova, Italy;

³Pediatric Neurology and Muscle Diseases Unit, IRCCS Istituto Giannina Gaslini, Genova, Italy, ⁴Radiology Unit, IRCCS Istituto Giannina Gaslini, Genova, Italy

PLENARY ROOM

17:42 - 17:54 MAPPING DISEASE PROGRESSION AND NEUROCOGNITION IN A SINGLE CENTRE COHORT OF LONG-LIVING ADULTS WITH DUCHENNE MUSCULAR DYSTROPHY (DMD): THE ROLE OF PLASMATIC BIOMARKERS OF NEURODEGENERATION

Gadaleta G.¹, Batti V.², Urbano G.¹, Vercelli L.¹, Rolle E.¹, Boschi S.¹, Leopizzi E.¹, Stura I.³, Gioiello G.⁴, Limoncelli S.⁴, Mengozi G.⁵, Geminiani G.C.⁶, Mongini T.E.¹

¹Neuromuscular Unit, Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin - Italy; ²Clinical Psychology Unit, A.O.U. Città della Salute e della Scienza di Torino, Turin - Italy; ³Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin - Italy; ⁴Clinical Biochemistry laboratory "Baldi e Riberi", A.O.U. Città della Salute e della Scienza di Torino, Turin - Italy; ⁵Clinical Biochemistry laboratory "Baldi e Riberi", A.O.U. Città della Salute e della Scienza di Torino; Department of Medical Sciences, University of Turin, Turin - Italy; ⁶Clinical Psychology Unit, A.O.U. Città della Salute e della Scienza di Torino; Department of Psychology, University of Turin, Turin - Italy

17:54 - 18:06 DIAPHRAGM ULTRASOUND AS BIOMARKER OF RESPIRATORY FUNCTION IN DUCHENNE MUSCULAR DYSTROPHY: A MULTICENTER STUDY

Trucco F.^{*1}, Ciceri L.², Roma E.³, Leone D.⁴, Lizio A.³, De Rose C.⁵, Buonsenso D.⁵, Albamonte E.³, Palermo C.⁴, Berti B.⁴, Becchiati S.³, De Gasperi M.³, Iatomasi M.³, Tarsia P.², Mercuri E.⁴, Pane M.⁴, Sansone V.³

¹Paediatric Neurology and Muscular Diseases Unit, Istituto di Ricovero e Cura a Carattere Scientifico Istituto Giannina Gaslini, Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Genova; ²Unit of Pulmonology, Ospedale Niguarda, Milan - Italy; ³NeMO Clinical Center, Fondazione Serena, Milan, Italy, ⁴Centro Clinico NeMO, Fondazione Policlinico Universitario Agostino Gemelli Istituto di Ricovero e Cura a Carattere Scientifico, Rome, Italy; ⁵Unit of Paediatrics, Dipartimento Scienze della salute della donna, del bambino e di sanità pubblica, Fondazione Policlinico Universitario Agostino Gemelli Istituto di Ricovero e Cura a Carattere Scientifico, Rome, Italy

18:06 - 18:18 PUTATIVE GENETIC MODIFIERS OF JOINT RANGE OF MOTION IN DUCHENNE MUSCULAR DYSTROPHY

Sabbatini D.¹, Gordish-Dressman H.², Duong T.³, Bello L.¹

¹Department of Neurosciences, DNS, University of Padova, Padova - Italy; ²Center for Genetic Medicine, Children's Research Institute, Children's National Health System, Washington, DC - United States of America; ³Department of Neurology, Stanford University School of Medicine, Palo Alto, CA - United States of America

18:18 - 18:30 VALIDATION OF 3D-MUSCLE ON CHIP PLATFORM FOR PATIENT ORIENTED PRECLINICAL STUDIES IN DUCHENNE MUSCULAR DYSTROPHY

Quarta R.¹, Cristiano E.¹, Han M.², Marinelli M.¹, Gaio N.², Mouly V.³, De Luca A.¹, Cappellari O.¹

¹University of Bari, Bari - Italy; ²BIOND Solutions B.V., Delft - Netherlands; ³Sorbonne Université, Institut de Myologie, Paris - France

PARALLEL ROOM 1

17:30 - 18:30

ORAL COMMUNICATION | MYASTHENIA GRAVIS

Chairpersons: Maurizio Inghilleri (Roma), Alessia Pugliese (Messina)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42

RAVULIZUMAB FOR GENERALIZED MYASTHENIA GRAVIS: A MULTICENTER REAL-LIFE EXPERIENCE

Rossini E.¹, Di Stefano V.², Iorio R.³, Habetswallner F.⁴, Maestri M.⁵, Vinciguerra C.⁶, Pennisi E.M.⁷, Di Martino G.², Rini N.², Falso S.³, Marini S.³, Ricciardi D.⁴, Guida M.⁵, Morino S.⁸, Garibaldi M.¹, Leonardi L.⁸, Marando D.¹, Tufano L.¹, Antonini G.¹, Fionda L.⁸

¹Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Rome, Italy; ²Biomedicine, Neuroscience and Advanced Diagnostic (BIND) Department, University of Palermo, Palermo, Italy; ³Department of Neuroscience, Università Cattolica del Sacro Cuore, Rome, Italy; ⁴UOC Neuropsychopathology, AORN Cardarelli, Via Antonio Cardarelli 9, Naples - Italy; ⁵Neurology Unit, Department of Clinical & Experimental Medicine, University of Pisa, Pisa, Italy; ⁶Neurology Unit, University Hospital "San Giovanni di Dio e Ruggi d'Aragona", Department of Medicine Surgery and Dentistry, "Scuola Medica Salernitana", Neuroscience section, Salerno, Italy; ⁷Neuromuscular Diseases Center, Neurology Unit, San Filippo Neri Hospital, Rome, Italy; ⁸Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital, Rome, Italy

17:42 - 17:54

CLINICAL MEANING OF PROMS IN A LARGE COHORT OF MYASTHENIA GRAVIS PATIENTS

Cheli M.¹, Brusa L.², Bartolucci F.³, Mantegazza R.⁴, Gemma M.⁵, Pennoni F.², Maggi L.¹

¹Foundation IRCCS Neurological Institute Carlo Besta, Neuroimmunology and Neuromuscular Diseases Unit, Milan - Italy; ²University of Milano-Bicocca, Department of Statistics and Quantitative Methods, Milan - Italy; ³University of Perugia, Department of Economics, Perugia - Italy; ⁴Società italiana Miastenia, Milano - Italy; ⁵Foundation IRCCS Neurological Institute Carlo Besta, Milan - Italy

17:54 - 18:06

ITALIAN REAL-LIFE EVIDENCE OF ZILUCOPLAN IN GENERALIZED MYASTHENIA GRAVIS A MULTICENTRE STUDY

Di Stefano V.¹, Alboini P.E.², Alonge P.¹, Antozzi C.³, Bevilacqua L.⁴, Biasini F.⁵, Bisecco A.⁶, Bonanno S.³, Brighina F.¹, Codeluppi L.⁷, D'Alvano G.⁶, Damato V.⁸, Erra C.⁹, Fionda L.¹⁰, Florio L.², Guida M.¹¹, Habetswallner F.⁹, Iorio R.¹², Luppino P.¹, Maestri Tassoni M.¹¹, Maggi L.³, Marini M.¹², Marini S.¹², Morino S.¹⁰, Pugliese A.⁵, Rossini E.¹³, Rini N.¹, Saccà F.¹⁴, Sarnataro A.¹⁴, Tuccillo F.⁹, Vanoli F.³, Vinciguerra C.⁴, Frangiamore R.³

¹Department of Biomedicine, Neuroscience and Advanced Diagnostics (BiND), University of Palermo, Palermo - Italy; ²Neurology Unit, Fondazione IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo - Italy; ³Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan - Italy; ⁴Neurology Unit, Department of Medicine, Surgery and Dentistry "Scuola Medica Salernitana", University of Salerno, Salerno - Italy; ⁵Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy; ⁶Division of Neurology, Department of Advanced Medical and Surgical Sciences (DAMSS), University of Campania "Luigi Vanvitelli", Naples, Italy;

⁷Neurology Unit, Neuromotor & Rehabilitation Department, Azienda USL-IRCCS di Reggio Emilia, Reggio Emilia - Italy; ⁸Department of Neurosciences, Psychology, Drug Research and Child Health (NEUROFARBA), University of Florence, Florence - Italy; ⁹UOC Neurophysiopathology, AORN Cardarelli, Naples - Italy; ¹⁰Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital, Rome, Italy; ¹¹Neurology Unit, Department of Medical Specialties, Azienda Ospedaliero Universitaria Pisana, Pisa, Toscana, Italy; ¹²Neurology Unit, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy; ¹³Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Rome - Italy; ¹⁴Department of Neurological Sciences, Reproductive and Odontostomatological Sciences, University Federico II, Naples - Italy.

18:06 - 18:18 LONG TERM EFFECTIVENESS OF THYMECTOMY IN ANTI-ACHRAB LATE ONSET MYASTHENIA GRAVIS (LOMG): A RETROSPECTIVE CASE-CONTROL STUDY.

[Latini E.¹](#), [Guida M.¹](#), [Cepele A.¹](#), [Rizzo S.¹](#), [Iovino V.¹](#), [Zirafa C.²](#), [Romano G.²](#), [Aprile V.³](#), [Lucchi M.³](#), [Siciliano G.¹](#), [Ricciardi R.¹](#), [Maestri Tassoni M.¹](#)

¹Department of Clinical and Experimental Medicine, Neurology Unit, University of Pisa, Pisa - Italy, ²Minimally Invasive and Robotic Thoracic Surgery-Surgical, Medical, Molecular and Critical Care Pathology Department, University of Pisa, Pisa - Italy, ³Division of Thoracic Surgery, Department for Surgical, Medical, Molecular Pathology and Critical Care, University of Pisa, Pisa - Italy

18:18 - 18:30 MYASTHENIA GRAVIS AND PREGNANCY: CLINICAL EVALUATION AND THERAPEUTIC DATA FROM AN ITALIAN MULTICENTRE STUDY GROUP

[Pugliese A.¹](#), [Drago S.F.A.¹](#), [Guida M.²](#), [Maestri Tassoni M.²](#), [Frangiamore R.⁵](#), [Damato V.⁴](#), [Florio L.⁶](#), [Previtali S.C.⁷](#), [Sorrenti B.⁷](#), [Iorio R.⁸](#), [Grandis M.⁹](#), [Garnero M.⁹](#), [Fionda L.¹⁰](#), [Antonini G.¹⁰](#), [Maggi L.⁵](#), [Liguori R.¹¹](#), [Cheli M.⁵](#), [Alibrandi A.¹²](#), [Schiavolin M.⁴](#), [Cepele A.²](#), [Iovino V.²](#), [Biasini F.¹](#), [Barbaccia A.¹](#), [Alboini P.E.⁶](#), [Evoli A.⁸](#), [Antozzi C.³](#), [Rodolico C.¹](#)

¹Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy; ²Department of Clinical and Experimental Medicine, Neurology Unit, University of Pisa, Pisa - Italy; ³Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico C. Besta, Milano - Italy; ⁴Department of Neurosciences, Drugs and Child Health, University of Florence, Florence - Italy; ⁵Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico C. Besta, Milan - Italy; ⁶Neurology Unit, Fondazione IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo - Italy; ⁷Division of Neuroscience, Department of Neurology, Institute of Experimental Neurology, San Raffaele Scientific Institute, Milan - Italy; ⁸Neurology Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome - Italy; ⁹Department of Neuroscience, Ophthalmology and Genetics, Section of Neurology and Neurological Rehabilitation, University of Genoa, Genoa - Italy; ¹⁰Department of Neurosciences, Mental Health and Sensory Organs (NESMOS), Sapienza University of Rome, Rome - Italy; ¹¹Department of Biomedical and Neuromotor Sciences, University of Bologna, Bologna - Italy; ¹²Unit of Statistical and Mathematical Sciences, Department of Economics, University of Messina, Messina- Italy

PARALLEL ROOM 2

17:30 - 18:30

ORAL COMMUNICATION | METABOLIC MYOPATHIES

Chairpersons: Sabrina Ravaglia (Milano), Maurizio Moggio (Milano)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42

DEVELOPMENT OF AN IN-VITRO 3D MODEL FOR LATE-ONSET POMPE DISEASE AND PROOF-OF-CONCEPT OF A NOVEL CRISPR THERAPEUTIC STRATEGY

Torri F.¹, Lionello V.², Zinghirino F.³, Cavazza A.³, Benedetti S.³, Siciliano G.¹, Tedesco S.V.²

¹Department of Clinical and Experimental Medicine, University of Pisa, Italy; ²University College London, London, UK; Francis Crick Institute, London - UK; ³Zayed Center for Research, London - UK.

17:42 - 17:54

MAGNETIZATION TRANSFER IMAGING IN LATE ONSET POMPE DISEASE

Croce M.G.¹, Naz F.², Barzaghi L.², Paoletti M.³, Mongini T.E.⁴, Gasparini S.⁵, Filosto M.⁶, Maggi L.⁷, Sechi A.⁸, Grandis M.⁹, Sacchini M.¹⁰, Sciaccio M.¹¹, Bonizzoni C.³, Bergsland N.¹², Santini F.¹³, Deligianni X.¹³, Gandini Wheeler-Kingshott C.A.M.¹⁴, Ravaglia S.¹⁵, Pichiecchio A.¹

¹Department Of Brain And Behavioural Sciences, University Of Pavia - Italy; ²Department Of Mathematics, University Of Pavia, Pavia - Italy; ³Advanced Imaging And Artificial Intelligence Center, Neuroradiology Department, IRCCS Mondino Foundation, Pavia - Italy; ⁴Neuromuscular Unit, Department Of Neurosciences, University Of Turin, Turin - Italy; ⁵Department Of Pediatrics, Università Degli Studi Milano-Bicocca, Fondazione MBBM, San Gerardo Hospital, Monza - Italy; ⁶Department Of Clinical And Experimental Sciences, University Of Brescia, Brescia - Italy; ⁷Neuroimmunology And Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ⁸Regional Coordinator Center For Rare Diseases, Academic Hospital Of Udine, Udine - Italy; ⁹Università Di Genova, Genoa - Italy; ¹⁰Unit Of Hereditary Metabolic And Muscular Disorders, Meyer Children's University Hospital - Firenze - Italy; ¹¹IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular And Rare Disease Unit - Milan - Italy; ¹²Department Of Neurology, Jacobs School Of Medicine And Biomedical Sciences, Buffalo Neuroimaging Analysis Center, University Of Buffalo, The State University Of New York, Buffalo - United States of America; ¹³Department Of Radiology, University Hospital Basel, Basel - Switzerland; ¹⁴Queen Square MS Centre, Department of Neuroinflammation, UCL Institute of Neurology, Faculty of Brain Sciences, University College London - London - United Kingdom; ¹⁵IRCCS C. Mondino Foundation, Pavia - Italy

17:54 - 18:06

INCREASED FREQUENCY OF GAA VARIANTS IN UK BIOBANK SUGGESTS UNDERDIAGNOSIS OF LATE-ONSET POMPE DISEASE

Gagliardi D.¹, Villella C.², Corti S.², Comi G.P.², Tucci A.³, Houlden H.⁴, Ronchi D.²

¹IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milan, Italy; ²Dino Ferrari Centre, Department of Pathophysiology and Transplantation (DEPT), Università degli Studi di Milano, Milano - Italy; ³William Harvey Research Institute, Queen Mary University of London, London, United Kingdom, London - United Kingdom; ⁴Department of Neuromuscular Diseases, Institute of Neurology, University College London, London, United Kingdom, London - United Kingdom

18:06 - 18:18 MUSCLE MRI EARLY SIGN IN PRESYMPTOMATIC PATIENTS WITH LATE-ONSET POMPE DISEASE (LOPD)
Porcino M.¹, Cavallaro M.², Bannino M.C.¹, Arena I.¹, Usbergo C.¹, Rodolico C.¹, Granata F.², Toscano A.¹, Gaeta M.², Musumeci O.¹
¹University of Messina, Messina - Italy; ²Radiology Department AOU G. Martino, Messina - Italy

18:18 - 18:30 EVALUATION OF SINGLE BREATH COUNT TEST FOR EARLY DETECTION OF RESPIRATORY DYSFUNCTION IN LATE ONSET POMPE DISEASE: PRELIMINARY DATA FROM A SINGLE CENTER EXPERIENCE
Arena I.G.², Porcino M.¹, Bannino M.C.¹, Usbergo C.¹, Toscano A.¹, Musumeci O.¹
¹Clinical and Experimental Medicine Department, University of Messina, Messina - Italy; ²BIOMORF Department, University of Messina, Messina - Italy

PLENARY ROOM

18:30 - 19:15 **Symposium Sponsored by SANTHERA | NO CME**
Chairperson: *Grazia D'Angelo (Milano)*
Vamorolone: a new therapeutic perspective in DMD

18:30 - 18:40 Introduction and Clinical Context of DMD
Claudio Bruno (Genova)

18:40 - 18:50 Vamorolone: Mechanism of Action and Clinical Implications
Luca Bello (Padova)

18:50 - 19:05 Vamorolone in DMD: Clinical Evidence and Therapeutic Perspectives
Luca Bello (Padova)

19:05 - 19:15 Discussion and Q&A

19:15 - 19:45 **Lecture sponsored by NOVARTIS | NO CME**
Chairperson: *Sonia Messina (Messina)*

Onasemnogene abeparvovec and new phenotypes of Spinal Muscular Atrophy: long-term data of patients treated with gene therapy
Riccardo Masson (Milano)

19:45 - 21:00 **Welcome Cocktail**

PLENARY ROOM

08:00 - 08:30 **Lecture 1**

Chairperson: *Rocco Liguori (Bologna)*

Immunotherapy in MG from Multi-Target to Target-Specific Therapies

Giovanni Antonini (Roma)

Parallel Workshop 1

Chairpersons: *Lorenzo Maggi (Milano), Massimiliano Mirabella (Roma)*

Update on myositis

08:30 - 08:50 New antibodies in myositis

Olivier Benveniste (Paris)

08:50 - 09:10 New pathological mechanisms in myositis

Kanneboyina Nagaraju (Binghamton)

09:10 - 09:30 Update on Sporadic Late Onset Nemaline Myopathy (SLONM)

Matteo Garibaldi (Roma)

09:30 - 10:00 Discussion and Q&A

PARALLEL ROOM 1

Parallel Workshop 2

Chairperson: *Adele D'Amico (Roma)*

Congenital myopathies

08:30 - 08:50 Laminopathies

Giovanna Lattanzi (Bologna)

08:50 - 09:10 Titin

Marco Savarese (Helsinki)

09:10 - 09:30 Collagen VI myopathies: from pathomolecular mechanisms to prospective therapies

Paolo Bonaldo (Padova)

09:30 - 10:00 Discussion and Q&A

PLENARY ROOM

10:00 - 10:30 **Lecture 2**

Chairperson: *Michelangelo Mancuso (Pisa)*

Phenotypes and treatments of TK2 deficiency

Cristina Dominguez (Madrid)

10:30 - 11:00 Coffee Break

PLENARY ROOM

Plenary Workshop 2

Chairperson: *Michelangelo Mancuso (Pisa)*

Controversies in Mitochondrial Diseases: Debates and Emerging Perspectives

11:00 - 11:20 **Lecture 1:**
Very Low Levels of Heteroplasmy: Clinical Relevance or Incidental Finding?
Amel Karaa (Paris)

11:20 - 11:40 **Lecture 2:**
Muscle Biopsy in Mitochondrial Diseases: Still Needed in the Genomic Era?
Olimpia Musumeci (Messina)

11:40 - 12:00 **Lecture 3:**
The Use of Mitochondrial Vitamin Cocktails: Scientific Evidence vs. Clinical Practice
Marcello Bellusci (Madrid)

12:00 - 12:20 **Lecture 4:**
Emerging treatments: gene editing vs metabolic-driven mitochondrial biogenesis
Mickal Minzuck (Cambridge)

12:20 - 12:30 Discussion and Q&A

12:30 - 13:30 Lunch and GDS Meetings

Myasthenia Gravis | **PLENARY ROOM**

Mitochondrial Disorders | **PARALLEL ROOM 1**

LGMD (Limb-Girdle Muscular Dystrophies) | **PARALLEL ROOM 2**

PARALLEL ROOM 1

13:30 - 14:15 **Symposium Sponsored by ROCHE | NO CME**

Chairperson: *Stefania Corti (Milano)*

Neuromuscular diseases from trials to clinical practice: what we've learned

13:30 - 13:35 Introduction
Stefania Corti (Milano)

13:35 - 13:50 Focus on SMA
Giorgia Coratti (Roma)

13:50 - 14:05 Focus on DMD
Eugenio Maria Mercuri (Roma)

14:05 - 14:15 Recap and Q&A

14:15 - 15:00 **Symposium Sponsored by BIOGEN | NO CME**

Chairperson: *Sonia Messina (Messina)*

Advancement in SMA: what do Nusinersen real world data suggest and what are the future perspectives?

14:15 - 14:20 Welcome and introduction
Sonia Messina (Messina)

14:20 - 14:40 New natural history in SMA: from real-life data to individualized trajectories of disease
Eugenio Maria Mercuri (Roma)

14:40 - 15:00 How clinical studies are filling the gaps and meeting evolving needs in SMA
Valeria Sansone (Milano)

15:00 - 15:45 **Symposium Sponsored by DYNE THERAPEUTICS | NO CME**

Chairperson: *Eugenio Maria Mercuri (Roma)*

Harnessing the FORCETM platform to advance targeted therapies for neuromuscular diseases

15:00 - 15:05 Welcome
Eugenio Maria Mercuri (Roma)

15:05 - 15:20 The clinical impact of the FORCE platform: Duchenne muscular dystrophy (DMD)
Marika Pane (Roma)

15:20 - 15:35 The clinical impact of the FORCE platform: myotonic dystrophy type 1 (DM1)
Valeria Sansone (Milano)

15:35 - 15:40 Q&A and panel discussion - *All faculty*

15:40 - 15:45 Closing
Eugenio Maria Mercuri (Roma)

15:45 - 16:15 Coffee Break

PLENARY ROOM

16:15 - 17:30 ORAL COMMUNICATION | MYASTHENIA GRAVIS Chairpersons: Elena Saccani (Parma), Amelia Evoli (Roma)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

16:15 - 16:27 THE ROLE OF WEARABLE INERTIAL MEASUREMENT UNITS (IMUS) TO ASSESS MOTOR PERFORMANCE IN PATIENTS WITH JUVENILE MYASTHENIA GRAVIS (JMG): A PILOT STUDY

Corsini C.¹, Fetta A.¹, Bisi M.C.², Di Pisa V.¹, Utili A.¹, Schiavo F.¹, Soliani L.¹, Pini A.¹, Giannotta M.¹, Stagni R.², Cordelli D.M.¹

¹IRCCS Institute of Neurological Sciences of Bologna, Pediatric Neuropsychiatry Unit, Bologna - Italy; ²University of Bologna, Department of Electric Electronic and Information Engineering "Guglielmo Marconi" - DEI, Bologna - Italy

16:27 - 16:39 ACCURACY OF ANTIBODY TESTING IN MYASTHENIA GRAVIS

Giannoccaro M.P.¹, Serra L.³, Grondona A.G.², Ricciardiello F.³, Ballestracci C.⁴, De Pasqua S.¹, Fetta A.⁵, Corsini C.⁵, Rossi S.⁶, Kaveh Moghadam K.⁷, Zini A.⁷, Rinaldi R.⁶, Cordelli D.M.⁵, Liguori R.¹

¹IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Clinica Neurologica, Bologna - Italy; ²Laboratorio Unico Metropolitano (LUM), Ospedale Maggiore, Bologna - Italy; ³IRCCS Istituto delle Scienze Neurologiche di Bologna, Laboratorio di Patologia Neuromuscolare e Neuroimmunologia, Bologna - Italy; ⁴Dipartimento di Scienze Biomediche e Neuromotorie, Università di Bologna, Bologna - Italy; ⁵IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Neuropsichiatria dell'Età Pediatrica - Bologna - Italy; ⁶IRCCS Istituto Delle Scienze Neurologiche Di Bologna, UOC Clinica Neurologica - Rete Neurologica Metropolitana (NEUROMET), Bologna - Italy; ⁷IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Neurologia e Rete Stroke Metropolitana, Ospedale Maggiore, Bologna - Italy

16:39 - 16:52 SPORADIC VERSUS IMMUNE CHECKPOINT INHIBITORS-ASSOCIATED MYASTHENIA GRAVIS-INFLAMMATORY MYOPATHY: A COMPARATIVE ANALYSIS

Rossini E.¹, Lauletta A.¹, Rossi S.², Damato V.³, Vogrig A.⁴, Marchioni E.⁵, Petrucchi A.⁶, Romano C.⁷, Lotti A.³, Florea I.⁴, Bini P.⁵, Tufano L.¹, Marando D.¹, Leonardi L.⁸, Morino S.⁸, Antonini G.¹, Garibaldi M.¹, Fionda L.⁸

¹Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Rome - Italy; ²IRCCS Istituto delle Scienze Neurologiche di Bologna, Bologna, Italy, Bologna - Italy; ³Department of Neurosciences, Psychology, Drug Research and Child Health (NEUROFARBA), University of Florence, Florence - Italy; ⁴Clinical Neurology, Department of Head-Neck and Neuroscience, Azienda Sanitaria Universitaria Friuli Centrale (ASU FC), Udine - Italy; Department of Medicine (DMED), University of Udine, Udine - Italy; ⁵Unit of Neurooncology, Fondazione IRCCS Mondino, Pavia - Italy; ⁶Center for Neuromuscular and Neurological Rare Diseases, San Camillo Forlanini Hospital, Rome - Italy; ⁷Stroke unit, Department of emergency, Ospedale dei Castelli-ASL6, Rome, - Italy; ⁸Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital, Rome - Italy

16:52 - 17:04 ESTABLISHING STABLE ACETYLCHOLINE RECEPTOR EXPRESSION IN RD CELLS USING THE PIGGYBAC SYSTEM: IMPROVING PLATFORMS FOR FUNCTIONAL ASSAYS

Morcone V.¹, Morroni J.², Valenzano G.¹, Marini S.², Falso S.², Marini M.², Iorio R.²

¹Università Cattolica del Sacro Cuore, Roma - Ital; ²Policlinico Universitario Agostino Gemelli IRCCS, Roma - Italy

17:04 - 17:16 REFRACTORY MYASTHENIA GRAVIS - CLINICAL COURSE AND PREDICTIVE FACTORS IN A LARGE COHORT OF PATIENTS

*Giacopuzzi Grigoli E.^{*1}, Calvacante P.¹, Frangiamore R.¹, Bonanno S.¹, Vanoli F.¹, Cheli M.¹, Elkadia M.², Salvi E.², Baggi F.¹, Andreetta F.¹, Canciello A.¹, Mantegazza R.¹, Antozzi C.¹, Maggi L.¹*

¹Neuroimmunology and Neuromuscular Disease Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan - Italy; ²Data Science Center, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano - Italy

17:16 - 17:30 THE EVOLVING LANDSCAPE OF MUSK EPIDEMIOLOGY AND TREATMENT

Falso S.¹, Marini S.¹, Valenzano G.¹, Morcone V.¹, Morroni J.², Marini M.¹, Habetswallner F.¹, Evoli A.², Iorio R.²

¹Department of Neuroscience, Catholic University of the Sacred Heart, Rome - Italy;

²Neurology Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome - Italy

PARALLEL ROOM 1**16:15 - 17:30 ORAL COMMUNICATION | MUSCULAR DYSTROPHIES**

Chairpersons: Paola Tonin (Verona), Maria Sframeli (Messina)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

16:15 - 16:27 MULTIDISCIPLINARY SCREENING FOR A NOVEL LMNA FOUNDER MUTATION IN A 2,948-MEMBER FAMILY TREE: A CITIZEN SCIENCE MODEL OF CARE

Peretto G.¹, Villatore A.¹, Bosco L.², Porro P.³, Strano C.M.M.², Sorrenti B.², Sista A.⁴, Ceres P.⁴, Mocetti C.³, Cecere A.³, Cione G.⁵, Carrera P.⁶, Esposito A.⁷, Della Bella P.⁸, Previtali S.C.², Di Resta C.⁶, Sala S.¹

¹Disease Unit for Myocarditis and Arrhythmogenic Cardiomyopathies, IRCCS San Raffaele Scientific Institute, Milan - Italy; ²Institute of Experimental Neurology (INSPE), Division of Neuroscience, IRCCS San Raffaele Scientific Institute, Milan - Italy; ³Laife Reply, Milan - Italy;

⁴Civil society of Caposele Caposele Italy; ⁵Avellino Azienda Sanitaria Locale (ASL), Avellino Italy; ⁶Genomic Unit for the Diagnosis of Human Pathologies, IRCCS San Raffaele Scientific Institute, Milan - Italy; ⁷Experimental Imaging Center, Radiology Unit, IRCCS San Raffaele Scientific Institute, Milan - Italy; ⁸Cardiac Electrophysiology Department, IRCCS San Raffaele Scientific Institute, Milan - Italy

16:27 - 16:39 SOLVING UNSOLVED NEUROMUSCULAR DISEASES BY WHOLE GENOME SEQUENCING

Zanobio M.¹, Di Letto P.¹, Palmieri F.P.¹, Cesarone E.², Picillo E.², Scarpato M.¹, Torella A.¹, Vicidomini G.¹, Giorgio E.³, Trapani I.⁴, Iovino E.⁵, Pippucci T.⁵, Nigro V.¹, Piluso G.¹

¹Department of Precision Medicine, University of Campania "Luigi Vanvitelli", Napoli - Italy;

²Medical Genetics and Cardiomyology, AOU Luigi Vanvitelli, Napoli - Italy; ³Department of Molecular Medicine, University of Pavia, Pavia - Italy; ⁴Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli - Italy; ⁵Medical Genetics Unit, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna - Italy

16:39 - 16:52 BETHLEM MYOPATHY AND INTRACRANIAL ANEURYSMS, SEGREGATION IN A FAMILY CARRYING A COL6A3 GENE MUTATION

Auletta M.¹, Sanricca C.², Primiano G.A.³, Servidei S.⁴

¹Dipartimento Universitario di Neuroscienze, Università Cattolica del Sacro Cuore Roma Italy, ²Neurophysiopathology unit Fondazione Policlinico Universitario A. Gemelli Roma and Fondazione UILDM Lazio Rome Italy, ³Neurophysiopathology unit Fondazione Policlinico Universitario A. Gemelli Roma Roma Italy, ⁴Dipartimento Universitario di Neuroscienze, Università Cattolica del Sacro Cuore, Neurophysiopathology unit Fondazione Policlinico Universitario A. Gemelli Roma Italy

16:52 - 17:04 COMPUTATIONAL MODELS FOR NEW PATIENT STRATIFICATION STRATEGIES OF NEUROMUSCULAR DISORDERS: THE COMPASS-NMD PROJECT

Nuredini A.¹, Socha M.², Suwalska A.², Pini S.¹, Corrias R.³, Dreyling C.⁴, Topf A.⁵, Vandroux M.⁶, Verdú Díaz J.⁵, Garzo Manzanera A.⁷, Albano N.¹, Saak A.⁴, Laporte J.⁶, Diaz Manera J.⁵, Obach M.⁷, Scipioni M.P.³, Savarese M.⁸, Santorelli F.M.⁹, Straub V.⁵, Schoser B.⁴, Polanska J.², Tupler R.G.¹

¹Department of Biomedical, Metabolic and Neural Sciences, University of Modena and Reggio Emilia, Modena - Italy; ²Department of Data Science and Engineering, Silesian University of Technology, Gliwice - Poland; ³Fincons Group, Lugano - Switzerland; ⁴Friedrich-Baur-Institut, Department of Neurology, Ludwig Maximilian University (LMU) Munich, Germany; ⁵John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle Hospitals NHS Foundation Trust, Newcastle University, Newcastle Upon Tyne - United Kingdom; ⁶Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC), Université de Strasbourg, Illkirch - France, ⁷Tecnalia Research and Innovation, San Sebastian - Spain; ⁸Folkhälsan Research Center, Helsinki - Finland; ⁹Molecular Medicine for Neurodegenerative and Neuromuscular Diseases Unit, IRCCS Fondazione Stella Maris, Pisa - Italy

17:04 - 17:16 FEVER-INDUCED RHABDOMYOLYSIS IN A 15-YEAR OLD BOY WITH HETEROZYGOUS PYGM AND DAG1 VARIANTS

Sogus E.¹, Vianello S.¹, Capece G.¹, Riguzzi P.¹, Petrosino A.¹, Zuin A.², Gueraldi D.², Sartori S.², Toldo I.², Bertolin C.², Salviati L.², Pegoraro E.¹, Bello L.¹

¹Department of Neurosciences, DNS, University of Padova, Padova - Italy, ²Department of Women's and Children's Health, University of Padua, Padua - Italy

17:16 - 17:30 CLINICAL AND MOLECULAR CHARACTERIZATION OF DUCHENNE MUSCULAR DYSTROPHY PATIENTS HARBOURING EXON 2 DUPLICATION

Zambon A.¹, Mietto M.², Canessa E.C.³, Ojo B.K.³, Falzarano M.S.², Fiorillo C.⁴, D'Amico A.⁵, Palermo C.⁶, Pane M.⁶, Urbano G.¹⁹, Ricci F.⁷, Allegra C.⁸, Messina S.⁸, Bello L.⁹, Pegoraro E.⁹, D'Angelo M.G.¹⁰, Masson R.¹¹, Briganti E.¹², Pini A.¹², Albamonte E.¹³, Picillo E.¹⁴, Nigro V.¹⁴, Astrea G.¹⁵, Magri F.¹⁶, Salvatici R.², Mazza T.¹⁸, Agolini E.¹⁸, Novelli A.¹⁸, Hathout Y.³, Ferlini A.², Previtali S.¹⁷

¹IRCCS Ospedale San Raffaele, Milano - Italy, ²Unit of Medical Genetics, Department of Medical Sciences, University of Ferrara, Ferrara - Italy; ³School of Pharmacy and Pharmaceutical Sciences, Binghamton University, New York - United States of America, ⁴Unit of Child Neuropsychiatry, IRCCS Istituto Giannina Gaslini and DINOGMI, UNIVERSITY of Genova, Genova - Italy; ⁵Unit of Muscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS, Rome - Italy; ⁶Centro Clinico NeMO, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome - Italy; ⁷Department of Sciences of Public Health and Paediatrics, University of Turin, Turin - Italy; ⁸Department of Clinical

and Experimental Medicine, University of Messina, Messina - Italy; ⁹Neurology Unit, Azienda Ospedale Padova Università Padova, Padua - Italy; ¹⁰Unit of Rare Diseases of the Central and Peripheral Nervous System Scientific Institute IRCCS E Medea, Bosisio Parini - Italy; ¹¹Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano - Italy; ¹²Child Neurology and Psychiatry Unit, IRCCS Istituto Delle Scienze Neurologiche Di Bologna, Bologna - Italy; ¹³The NEMO Clinical Center in Milan, Neurorehabilitation Unit, University of Milan, ASST Niguarda Hospital, Milano - Italy; ¹⁴Medical Genetics and Cardiomyology Unit, Department of Precision Medicine, Università Degli Studi Della Campania "Luigi Vanvitelli", Naples, Italy and TIGEM, Pozzuoli - Italy; ¹⁵Department of Neuroscience, IRCCS Fondazione Stella Maris, Pisa - Italy; ¹⁶Neuromuscular and Rare Diseases Unit, Department of Neuroscience, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan - Italy; ¹⁷Università Vita e Salute San Raffaele, Milano - Italy; ¹⁸Laboratory of Medical Genetics, IRCCS Ospedale Pediatrico Bambino Gesù, Rome Italy; ¹⁹Neuromuscular Unit, Department of Neurosciences RLM, University of Torino, Torino Italy

PARALLEL ROOM 2

16:15 - 17:30 **ORAL COMMUNICATION: EXPERIMENTAL THERAPIES, MITOCHONDRIAL DISORDERS, CONGENITAL MYOPATHIES** Chairpersons: *Daria Diodato (Roma), Dario Ronchi (Milano)*

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

16:15 - 16:27 **THYMIDINE KINASE 2 DEFICIENCY: A POTENTIALLY TREATABLE AND UNDERESTIMATED MYOPATHY**

Pettenuzzo J.⁸, Pelle S.¹, Diodato D.², Procopio E.³, Bello L.⁴, Pegoraro E.⁴, Musumeci O.⁵, Ronchi D.⁶, Mancuso M.⁷, Garone C.⁸

¹Department of Medical and Surgical Sciences, Alma Mater Studiorum - University of Bologna, Bologna - Italy; ²Neuromuscular Disorders Research Unit, Bambino Gesù Children's Hospital, IRCCS, Roma - Italy; ³Metabolic and Muscular Unit, Meyer Children's Hospital IRCCS, Firenze - Italy; ⁴Department of Neurosciences, Motor Neuron Disease Center, University Hospital of Padova, Padova - Italy; ⁵Department of Clinical and Experimental Medicine, University of Messina - Unit of Neurology and Neuromuscular Disorders AOU G. Martino, Messina - Italy; ⁶Dino Ferrari Center, Department of Pathophysiology and Transplantation, University of Milan - IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milano - Italy; ⁷Neurological Institute, Department of Clinical and Experimental Medicine, University of Pisa, Pisa - Italy; ⁸Department of Medical and Surgical Sciences, Alma Mater Studiorum - University of Bologna - IRCCS Istituto delle Scienze Neurologiche di Bologna, U.O.C. Neuropsichiatria dell'età pediatrica, Bologna - Italy

16:27 - 16:39 AUTOMATING DNA MUTATION IDENTIFICATION IN PRIMARY MITOCHONDRIAL DISEASES USING NON-GENETIC AND HISTOLOGICAL-FREE CLINICAL DATA

Mazzucato S.², Lopriore P.¹, Pierotti F.², Cecchi G.¹, Lasala A.², Lamperti C.³, Einvaig K.³, Nicoletta V.³, Carelli V.⁴, Lamorgia C.⁴, Valentino M.L.⁴, Musumeci O.⁵, Arena I.G.⁵, Servidei S.⁶, Primiano G.⁶, Mongini T.⁷, Urbano G.⁷, Procopio E.⁸, Ruggiero L.⁹, Diodato D.¹⁰, Bertini E.¹⁰, Garone C.¹¹, Rinaldi R.¹², Montano V.¹, Siciliano G.¹, Micera S.², Mancuso M.¹, Bandini A.²

¹Department of Clinical and Experimental Medicine, Neurological Clinic, University of Pisa, Pisa - Italy; ²Biorobotics Institute, Department of Excellence in Robotics and AI - Scuola Superiore Sant'Anna, Pisa - Italy; ³UO Medical Genetics and Neurogenetics, Fondazione IRCCS Istituto Neurologico C. Besta, Milan - Italy; ⁴IRCCS Istituto delle Scienze Neurologiche di Bologna, Programma di Neurogenetica, Bologna - Italy; ⁵Department of Clinical and Experimental Medicine, UOC Neurologia e Malattie Neuromuscolari, University of Messina, Messina - Italy; ⁶Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome - Italy; ⁷Neuromuscular Unit, Department of Neurosciences, University of Torino, Turin - Italy; ⁸Metabolic Unit, Meyer Children's Hospital IRCCS, Florence - Italy; ⁹Dipartimento di Neuroscienze e Scienze Riproduttive ed Odontostomatologiche, Università degli Studi di Napoli "Federico II, Naples - Italy; ¹⁰Unit of Muscular and Neurodegenerative Disorders, Ospedale Pediatrico Bambino Gesù, Roma - Italy; ¹¹Department of Medical and Surgical Sciences, Alma Mater Studiorum University of Bologna, Bologna - Italy; ¹²IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Interaziendale Metropolitana (NeuroMet), - Neurologia AOU S.Orsola-Malpighi, Bologna - Italy

16:39 - 16:52 NEW PRECLINICAL THERAPIES FOR MITOCHONDRIAL MYOPATHIES

Zeviani M.¹⁻², Balmaceda V.³⁻⁴, Giacchin G.³⁻⁴, Cerutti R.³⁻⁴, Volta S.³⁻⁴, Viscomi C.³⁻⁴,

¹IRCCS Women and Children's Hospital "Burlo-Garofolo"; ² Department of Neurosciences, University of Padova, Padova - Italy; ³ Department of Biomedical Sciences, University of Padova, Padova - Italy; ⁴ Veneto Institute of Molecular Medicine (VIMM), Padova - Italy

16:52 - 17:04 LOW GLUCOSE KETOGENIC DIET IN MCARDLE DISEASE: A FATTY WAY FOR A STARVING MUSCLE

Martinuzzi A.¹, Musumeci O.², Stefan C.³, Vinante E.³, Perillo C.³, Psenti N.³, Toscano A.⁴

¹University of Turin, Turin - Italy; ² Unit of Neurology and Neuromuscular Disorders, Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy; ³IRCCS E.Medea, Department of Conegliano, Conegliano - Italy; ⁴ ERN-NMD Center for Neuromuscular Disorders of Messina, Department of Clinical and Experimental Medicine, AOU G. Martino of Messina, Messina - Italy

17:04 - 17:16 ENDOPLASMIC RETICULUM STRESS IN SELENOPROTEIN N1-RELATED MYOPATHY: A PROSPECTIVE ON THE DRUGGABILITY

Zito E.², Germani S.¹, Renna G.¹, Varone E.¹, Blaauw B.⁵, D'Amico A.³, Previtali S.⁴, Ferreiro A.⁶

¹Istituto Farmacologico Mario Negri, Milano - Italy; ²Università di Urbino Carlo Bo, Urbino - Italy; ³IRCCS, Bambino Gesù Children's Hospital, Rome - Italy; ⁴IRCCS Ospedale San Raffaele and University Vita-Salute San Raffaele, Milano - Italy; ⁵Università di Padova, Padova Italy; ⁶Sorbonne Université & Reference Center for Neuromuscular Disorders Nord-Est-Ile de France, Neuromyology Department, Groupe Hospitalier Pitie-Salpêtrière, Paris - France

**17:16 - 17:30 CONGENITAL HEART DEFECTS IN RECESSIVE TITINOPATHIES:
A PRELIMINARY STUDY**

Di Feo M.F.¹, Kurbatov S.², Attie-Bitach T.³, Marzin P.³, Savarese M.¹, Udd B.¹

¹Folkhälsan Research Centre Helsinki Finland; ²Voronezh State Medical University N.N. Burdenko, Voronezh - Russian Federation; ³Necker Hospital, Service de Médecine Génomique des Maladies Rares, Paris - France

PLENARY ROOM

Parallel Workshop 3

Chairperson: *Marina Grandis (Genova)*

Pregnancy and childbirth with neuromuscular diseases

17:30 - 17:50 Pregnancy and childbirth in Myasthenia Gravis

Carmelo Rodolico (Messina)

17:50 - 18:10 Pregnancy and childbirth in Myotonic Dystrophies

Valeria Sansone (Milano)

18:10 - 18:30 Pregnancy and childbirth in SMA

Lucia Ruggiero (Napoli)

18:30 - 19:00 Discussion and Q&A

PARALLEL ROOM 1

Parallel Workshop 4

Chairpersons: *Tiziana Mongini (Torino), Enzo Ricci (Roma)*

FSHD: Approaches to Clinical Diagnosis and Phenotyping

17:30 - 17:50 Towards an Italian registry for FSHD

Massimiliano Filosto (Brescia)

17:50 - 18:10 The revised CCEF for describing phenotypes

Giulia Ricci (Pisa)

18:10 - 18:30 Natural history and disease progression

Liliana Vercelli (Torino)

18:30 - 19:00 Discussion and Q&A

Friday, May 30, 2025

Friday, May 30, 2025

PARALLEL ROOM 1

- 08:30 - 09:00 **Breakfast Seminar Sponsored by ITALFARMACO | NO CME**
Chairperson: *Sonia Messina (Messina)*
Endpoints in Duchenne muscular dystrophy: from trials to clinical practice
- 08:30 - 08:40 Assessing milestone and disease progression in Duchenne muscular dystrophy
Marika Pane (Roma)
- 08:40 - 08:50 Translating trials endpoints into clinical practice
Luca Bello (Padova)
- 08:50 - 09:00 Closing remarks

PLENARY ROOM

Parallel Workshop 5

Chairperson: *Giorgio Tasca (Newcastle)*
MRI in muscular disorders

- 09:00 - 09:20 MRI in Muscle disorders
Giorgio Tasca (Newcastle)
- 09:20 - 09:40 MRI for diagnosis and monitoring in FSHD
Mauro Monforte (Roma)
- 09:40 - 10:00 AI in MRI
Josè Verdu Diaz (Newcastle)
- 10:00 - 10:30 Discussion and Q&A

PARALLEL ROOM 1

Parallel Workshop 6

Chairperson: *Antonio Trabacca (Brindisi), Lucio Santoro (Napoli)*
Emergencies in Neuromuscular Disorders

- 09:00 - 09:20 Neuromuscular disorders in an adult emergency department
Tiziana Mongini (Torino)
- 09:20 - 09:40 Neuromuscular disorders in a pediatric emergency department
Adele D'Amico (Roma)
- 09:40 - 10:00 Emergencies cards
Fabrizio Racca (Torino)
- 10:00 - 10:30 Discussion and Q&A
- 10:30 - 11:00 Coffee Break

PLENARY ROOM

Plenary Workshop 7

Chairperson: *Giacomo Pietro Comi (Milano)*

New therapeutic roads in muscle disorders

- 11:00 - 11:20 Myotonic dystrophy
Benedikt Schoer (Monaco)
- 11:20 - 11:40 FSHD
Enrico Bugiardini (Londra)
- 11:40 - 12:00 New Therapeutic Options in Pompe Disease
Antonio Toscano (Messina)
- 12:00 - 12:20 Ketogenic diet in metabolic myopathies
Andrea Martinuzzi (Torino)
- 12:20 - 12:30 Discussion and Q&A

PARALLEL ROOM 1

Parallel Workshop 8

Chairperson: *Claudio Bruno (Genova)*

The Role of Calcium in Neuromuscular Disorders

- 11:00 - 11:20 Calcium handling in congenital myopathies
Ester Zito (Milano)
- 11:20 - 11:40 New insights into the role of store operated calcium entry as a promising pharmacological target for Duchenne Muscular Dystrophy
Beatrice Riva (Novara)
- 11:40 - 12:00 The Mitochondrial Calcium Uniporter (MCU)
Rosario Rizzuto (Padova)
- 12:00 - 12:30 Discussion and Q&A
- 12:30 - 13:30 **Lunch and GDS Meetings**

Giovani AIM | **PLENARY ROOM**

FSHD (Facioscapulohumeral Muscular Dystrophy) | **PARALLEL ROOM 1**

Inflammatory Myopathies | **PARALLEL ROOM 2**

PARALLEL ROOM 1

13:30 - 14:15 **Symposium Sponsored by UCB | NO CME**

Chairperson: Carmelo Rodolico (Messina)

Myasthenia Gravis: connecting the Next-Generation C5 Inhibitor to Real Life

13:30 - 13:35 Opening Remarks
Carmelo Rodolico (Messina)

13:35 - 13:50 From Science to Practice: What makes zilucoplan different?
Rita Frangiamore (Milano)

13:50 - 14:05 Zeta Network: Italian Real-World Experience with zilucoplan
Vincenzo Di Stefano (Palermo)

14:05 - 14:15 Closing Remarks & Q&A
Carmelo Rodolico (Messina)

14:15 - 15:00 **Symposium Sponsored by ARGENX | NO CME**

Chairperson: Francesco Habetswallner (Napoli)

Pioneering innovation in personalized gMG treatment:
real world evidence in action

14:15 - 14:20 Welcome and introduction
Francesco Habetswallner (Napoli)

14:20 - 14:25 Achieving a Rapid and Deep Response in gMG: Insights from Clinical Practice
Yuri Falzone (Milano)

14:25 - 14:30 Sustaining Treatment Response Over Time: Long-Term Strategies in gMG
Laura Fionda (Roma)

14:30 - 14:35 Enhancing Quality of Life in gMG: Beyond Symptom Control
Rita Frangiamore (Milano)

14:35 - 14:40 Safety and Tolerability in gMG Treatment: Balancing Efficacy
and Patient Well Being
Elia Sechi (Sassari)

14:40 - 15:00 Discussion and Q&A - All

15:00 - 15:45 **Symposium Sponsored by ALEXION | NO CME**

Chairpersons: Carlo Antozzi (Milano), Francesco Habetswallner (Napoli)

Ravulizumab: innovation and Real-World Evidence sustaining Long-acting
efficacy in generalized Myasthenia Gravis

15:00 - 15:05 Introduction and Objectives:
Carlo Antozzi (Milano), Francesco Habetswallner (Napoli)

15:05 - 15:20 The added value of Long-acting C5 inhibition
Raffaele Iorio (Roma)

15:20 - 15:35 Optimizing treatment goals in gMG

Michelangelo Maestri (Pisa)

15:35 - 15:45 **Q&A and Discussion**

Carlo Antozzi (Milano), Francesco Habetswallner (Napoli), Raffaele Iorio (Roma), Michelangelo Maestri (Pisa)

15:45 - 17:30 **Coffee Break with Poster Session * NO CME pag. 35**

PLENARY ROOM

16:30 - 17:30 **MUSCLE TRIVIA AIM CONTEST | Qualification**

Chairpersons: *Claudio Bruno (Genova),
Serenella Servidei (Roma), Antonio Trabacca (Brindisi)*



PLENARY ROOM

17:30 - 18:30 ORAL COMMUNICATION | GENETICS

Chairpersons: *Francesca Magri (Milano), Filippo Santorelli (Pisa)*

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42 HOW TO HANDLE TITIN VARIANTS: ANALYSIS OF 18, 462 INDIVIDUALS FROM THE SOLVE-RD CONSORTIUM

Di Feo M.F.¹, Paramonov I.², Matalonga Borrel L.², Topf A.³, Hoischen A.⁴, Beltran S.², Graessner H.⁵, Vissers L.⁴, De Voer R.⁴, Van Gijn M.⁶, Balestrini S.⁷, Lerche H.⁸, Lesca G.⁹, Natraj Gayathri S.¹⁰, Ellwanger K.⁵, Cossee M.¹¹, Perrin A.¹¹, Sarkozy A.¹², Bonne G.¹³, Verdonschot J.¹⁴, Demidov G.¹⁵, Laurie S.², Johari M.¹⁶, Hackman P.¹⁰, Udd B.¹⁰, Savarese M.¹⁰

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17:42 - 17:54 LEVERAGING RNA SEQUENCING TO REVEAL THE MOLECULAR BASIS OF RARE NEUROMUSCULAR DISEASES

Lillback V.¹, Natraj Gayathri S.¹, Di Feo M.F.¹, Oghabian A.¹, Jonson P.H.¹, Lehtokari V.¹, Pelin K.¹, Johari M.², Hackman P.¹, Udd B.¹, Savarese M.¹

¹Myofin Research Group, Folkhalsan Research Center, Helsinki - Finland; ²Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Perth - Australia

17:54 - 18:06 A CASE SERIES OF LATE-ONSET MULTIPLE ACYL-COA DEHYDROGENASE DEFICIENCY DUE TO ETFDH GENE MUTATION WITH ACUTE-SUBACUTE ONSET

Zoppi D.¹, Russo A.¹, Vallefucio F.¹, De Maria M.¹, Boemia V.¹, Russo R.¹, Esposito G.², Santorelli F.M.³, Ruggiero L.¹

¹Department of Neurosciences, Reproductive Sciences, and Odontostomatology, University of Naples "Federico II", Napoli - Italy; ²CEINGE-Biotecnologie Avanzate s.c. a.r.l. / Department of Molecular Medicine and Medical Biotechnologies, University of Naples "Federico II", Napoli - Italy; ³Molecular Medicine, IRCCS Fondazione Stella Maris, Pisa - Italy

18:06 - 18:18 WHOLE EXOME SEQUENCING ANALYSIS IN INCLUSION BODY MYOSITIS
Velardo D.¹, Gagliardi D.², Magri F.¹, Zanotti S.¹, Piga D.², Napoli L.¹,
Ripolone M.¹, Ciscato P.¹, Bertolasi L.¹, Molitierno N.³, Fortunato F.³, Sciaccio M.¹,
Comi G.P.², Corti S.¹, Ronchi D.³

¹IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular Unit, Milan Italy; ²IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milan Italy; ³University of Milan, Dino Ferrari Center, Milan - Italy

18:18 - 18:30 COMMON ANCESTRY FROM THE SAME SMALL AREA OF SOUTHERN ITALY IS SUGGESTED BY TWO FAMILIES WITH DILATED CARDIOMYOPATHY WHO SHARE THE SAME HOMOZYGOUS LOSS-OF-FUNCTION VARIANT IN NRAP
Onore M.E.¹, Caiazza M.², Mio C.³, Borrelli R.N.¹, Monda E.², Scarano G.⁴,
Faletta F.⁵, Nigro V.⁶, Limongelli G.⁷, Piluso G.¹

¹Department of Precision Medicine, University of Campania "Luigi Vanvitelli", Naples - Italy; ²Inherited and Rare Cardiovascular Diseases Unit, Department of Translational Medical Sciences, University of Campania "Luigi Vanvitelli", Monaldi Hospital, Naples - Italy; ³Department of Medicine (DMED), University of Udine, Udine - Italy; ⁴Inherited and Rare Cardiovascular Diseases Unit, Department of Translational Medical Sciences, University of Campania "Luigi Vanvitelli", Monaldi Hospital; U.O.S.D. Genetica Medica, A.O.R.N. San Pio, Benevento, Naples - Italy; ⁵Institute of Medical Genetics, Azienda Sanitaria Universitaria Friuli Centrale (ASUFC), Udine - Italy; ⁶Department of Medicine (DMED), University of Udine Udine - Italy; ⁷Department of Precision Medicine, University of Campania "Luigi Vanvitelli"; Telethon Institute of Genetics and Medicine (TIGEM) Naples, Pozzuoli - Italy; ⁸Inherited and Rare Cardiovascular Diseases Unit, Department of Translational Medical Sciences, University of Campania "Luigi Vanvitelli", Monaldi Hospital; Institute of Cardiovascular Science, University College London and St. Bartholomew's Hospital Nap

PARALLEL ROOM 1

17:30 - 18:30 ORAL COMMUNICATION | SPINAL MUSCLE ATROPHY (SMA)
 Chairpersons: Antonella Pini (Bologna), Carlo Minetti (Genova)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42 UNLOCKING THE POWER OF CIRCULATING MYOMIRS AS BIOMARKERS FOR ADULT SMA PATIENTS UPON NUSINERSEN TREATMENT
Malacarne C.², Farinazzo G.², Zanin R.¹, Mehmeti E.³, Salvi E.⁴, Lipka J.⁵,
Hagenhacker T.⁵, Maggi L.², Marcuzzo S.⁶, Bonanno S.²

¹Developmental Neurology, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ²Neuroimmunology and Neuromuscular Disease Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ³Data Science Center, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ⁴Data Science Center, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ⁵Computational multi-Omics of Neurological Disorders (MIND) Lab, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy; ⁶Department of Neurology, and Center for Translational Neuro- and Behavioral Sciences (C-TNBS), University Medicine Essen, Essen - Germany; ⁷Neuroimmunology and Neuromuscular Disease Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan, Italy; Brain-targeted Nanotechnologies (BrainNs) Lab, Joint Research Platform, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan - Italy

17:42 - 17:54 NEUROPHYSIOLOGICAL BIOMARKERS IN ADULTS WITH TYPE III SPINAL MUSCULAR ATROPHY

[Libelli G.](#)¹, [Nuredini A.](#)¹, [D'Orsi A.](#)¹, [Allegri I.](#)², [Chierici E.](#)², [Saccani E.](#)²

¹Unit of Neurology, Department of Medicine and Surgery, University of Parma, Parma - Italy;

²Neurology Unit, Department of General and Specialized Medicine, University Hospital of Parma, Parma - Italy

17:54 - 18:06 BMI AND MOTOR FUNCTION IN A PEDIATRIC POPULATION OF ITALIAN TREATED PATIENTS WITH SMA: IS THERE A CORRELATION?

[Briganti E.](#)¹, [Not R.](#)¹, [Scarpini G.](#)¹, [Giannotta M.](#)¹, [Trentin F.](#)¹, [Parrinello G.](#)², [Baccari F.](#)³, [Duccio C.](#)⁴, [Coratti G.](#)⁵, [Agosto C.](#)⁶, [Bitetti I.](#)⁷, [Brolatti N.](#)⁸, [Bruno I.](#)⁹, [Coccia M.](#)¹⁰, [Filosto M.](#)¹¹, [Gagliardi D.](#)¹², [Maggi L.](#)¹³, [Messina S.](#)¹⁴, [Sframeli M.](#)¹⁴, [Nigro V.](#)¹⁵, [Pegoraro E.](#)¹⁶, [Ricci F.](#)¹⁷, [Sansone V.](#)¹⁸, [Siciliano G.](#)¹⁹, [Ticci C.](#)²⁰, [Trabacca A.](#)²¹, [Vacchiano V.](#)²², [Verriello L.](#)²³, [Mercuri E.](#)⁵, [Pini A.](#)¹

¹Pediatric Neuromuscular Unit, Pediatric Neuropsychiatry Unit, IRCCS Institute of Neurological Sciences, Bologna - Italy; ²School of Physical Therapy, University of Bologna, Bologna - Italy; ³Epidemiology and Statistics Unit, IRCCS Institute of Neurological Sciences Bologna - Italy; ⁴Department of Medical and Surgical Sciences, University of Bologna Bologna - Italy; ⁵Centro Clinico NeMO, Fondazione Agostino Gemelli IRCCS, Rome - Italy; ⁶Department of Women's and Children's Health, University of Padua, Padua - Italy; ⁷Pediatric Neurology, Neurology Unit, Santobono-Pausilipon Children's Hospital, Naples - Italy; ⁸Center of Translational and Experimental Myology, and Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, IRCCS Istituto Giannina Gaslini, Genoa - Italy; ⁹Institute for Maternal and Child Health, IRCCS Burlo Garofolo, Trieste - Italy; ¹⁰Department of Neurological Sciences, AOU Ospedali Riuniti di Ancona, Ancona - Italy; ¹¹Department of Clinical and Experimental Sciences, University of Brescia (Italy), NeMO-Brescia Clinical Center for Neuromuscular Diseases, Brescia - Italy; ¹²Pediatric Neurology Unit, Pediatric Hospital Giovanni XXIII, Bari - Italy; ¹³Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan - Italy; ¹⁴Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy; ¹⁵Medical Genetics and Cardiomyology Unit, Department of Precision Medicine, Università degli Studi della Campania Luigi Vanvitelli, Naples - Italy; ¹⁶Neurology Unit, Azienda Ospedale Padova, Padua - Italy; ¹⁷SC Neuropsichiatria Infantile, Dipartimento di Scienze della Sanità Pubblica e Pediatriche, Università di Torino, Turin - Italy; ¹⁸The NeMO Center in Milan, Neurorehabilitation Unit, University of Milan, ASST Niguarda Hospital, Milan - Italy; ¹⁹Neurology Unit, Department of Clinical and Experimental Medicine, AOU Pisana, Pisa - Italy; ²⁰Metabolic Unit, A. Meyer Children's Hospital, Florence - Italy; ²¹Developmental Neurology and Neurorehabilitation, Unit for Severe disabilities in Developmental Age and Young Adults, IRCCS "E. Medea", Brindisi - Italy; ²²UOC Clinica Neurologica, IRCCS Institute of Neurological Sciences, Bologna - Italy; ²³Neurology Unit, Department of Neurosciences, University Hospital Santa Maria della Misericordia, Udine - Italy

18:06 - 18:18 PROPOSAL FOR A NEW UPDATE NOMENCLATURE AND CLINICAL PROTOCOL MANAGEMENT OF SPINAL MUSCULAR ATROPHY IN THE ERA OF NEWBORN SCREENING

[Varone A.](#), [Bitetti I.](#)

Santobono Pausilipon, Napoli - Italy

18:18 - 18:30 NEWBORN SCREENING FOR SPINAL MUSCULAR ATROPHY (SMA) IN LOMBARDY: AN 18-MONTH REGIONAL MULTICENTER EXPERIENCE

Masson R.¹, Albamonte E.², Berardinelli A.³, Bonaventura E.⁴, Comi G.⁵, Corti S.⁵, Dosi C.¹, Filosto M.⁶, Lanteri P.¹, Mandelli A.⁴, Montrasio C.⁴, Parravicini S.¹, Sansone V.², Sommariva A.¹, Venerando A.¹, Zambon A.⁷, Gellera C.¹, Cereda C.⁴

¹Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano - Italy; ²The NEMO Center in Milan, University of Milan, ASST Niguarda Hospital, Milano - Italy; ³IRCCS C, Mondino Foundation, Pavia - Italy; ⁴Buzzi Children's Hospital, Milano - Italy; ⁵Foundation IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano - Italy; ⁶NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of clinical and Experimental Sciences, University of Brescia, Brescia - Italy; ⁷Unit of Neurology, IRCCS Ospedale San Raffaele, Milano - Italy

PARALLEL ROOM 2

17:30 - 18:30 ORAL COMMUNICATION |FSHD

Chairpersons: Rossella Tupler (Modena), Antonio Di Muzio (Chieti)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

17:30 - 17:42 FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY: UNCOVERING THE IMPACT OF 4Q35 GENES DYSREGULATION ON MUSCLE PROTEIN SYNTHESIS

Salsi V.¹, Losi F.¹, Fosso B.², Ferrarini M.¹, Pini S.¹, Manfredi M.⁶, Vattermi G.⁷, Mongini T.⁸, Maggi L.⁹, Pesole G.², Henras A.³, Kaufman P.⁵, Mcstay B.⁴, Tupler R.¹

¹University of Modena and Reggio Emilia, Modena - Italy; ²University of Bari Aldo Moro, Bari - Italy; ³Université de Toulouse, Toulouse - France; ⁴University of Galway, Galway - Ireland; ⁵University of Massachusetts, Worcester - United States of America; ⁶University of Piemonte Orientale, Novara - Italy; ⁷University of Verona - University of Verona, Verona - Italy; ⁸University of Turin, Torino - Italy; ⁹Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano - Italy

17:42 - 17:54 ANALYSIS OF GENETIC DIVERSITY AND PHYLOGENETIC RELATIONSHIPS OF D4Z4 REPEATS: IMPLICATION FOR HEALTH AND DISEASE

Chiara M.¹, Pini S.², Salsi V.², Horner D.¹, Pesole G.³, Tupler R.²

¹Department of Biosciences, Università degli Studi di Milano, Milan - Italy; ²Department of Biomedical, Metabolic and Neural Sciences, University of Modena and Reggio Emilia, Modena - Italy; ³Department of Biosciences, Biotechnology and Environment, University of Bari Aldo Moro, Bari - Italy

17:54 - 18:06 BIALLELIC PAX7 VARIANTS AS A CAUSE OF MYOPATHY WITH FSHD-LIKE CLINICAL FEATURES

Ganassi M.¹⁰, Strafella C.¹, Savarese M.², Heher P.³, Engquist E.N.³, McGuire L.³, Mridul J.², De Nicola G.F.³, Bigot A.⁴, Mouly V.⁵, Bortolani S.⁶, Torchia E.⁶, Monforte M.⁶, Megalizzi D.⁷, Sabino A.⁸, Ricci E.⁶, Giardina E.⁷, Zammit P.S.³, Tasca G.⁹

¹Genomic Medicine Laboratory UILDM, IRCCS Santa Lucia Foundation, Rome - Italy;

²Folkhälsan Research Center, Helsinki - Finland; ³King's College London, Randall Centre for Cell and Molecular Biophysics, London - United Kingdom; ⁴MyoLine Platform, Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris - France;

⁵UPMC University Paris 06, Inserm UMR5974, CNRS FRE3617, Center for Research in Myology, Sorbonne University, Paris - France; ⁶Unità Operativa Complessa di Neurologia, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome - Italy; ⁷Genomic Medicine Laboratory UILDM, IRCCS Santa Lucia Foundation; Department of Biomedicine and Prevention, Medical Genetics Laboratory, Tor Vergata University, Rome - Italy; ⁸Istituto di Neurologia, Università Cattolica del Sacro Cuore, Rome - Italy; ⁹John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trusts, Newcastle Upon Tyne - United Kingdom; ¹⁰King's College London, Randall Centre for Cell and Molecular Biophysics; Genetic Therapy Accelerator Centre, Queen Square Institute of Neurology, University College London, London - United Kingdom

18:06 - 18:18 ARTIFICIAL INTELLIGENCE FOR FSHD CARE: AN AI-POWERED ASSISTANT FOR PATIENTS AND HEALTHCARE PROVIDERS

Megalizzi D.⁶, Strafella C.¹, Trastulli G.¹, Proietti Piorgio E.¹, Colantoni L.¹, Torri F.², Risi B.³, Caltagirone C.⁴, Ricci G.², Siciliano G.², Filosto M.⁵, Cascella R.¹, Giardina E.⁶

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18:18 - 18:30 THE OVEREXPRESSION OF THE RNA BINDING PROTEIN FRG1 LEADS TO REDUCED MATURATION AND DECREASED METABOLIC EFFICIENCY IN SKELETAL MUSCLE

Fantini S.¹, Bisceglia G.¹, Zyla J.², Moio N.¹, Nuredini A.¹, D'Antona G.⁴, Vattemi G.³, Polanska J.², Tupler R.¹

¹Department of Biomedical, Metabolic and Neural Sciences University of Modena and Reggio Emilia, Modena - Italy; ²Department of Data Science and Engineering, Silesian University of Technology, 44-100 Gliwice, Poland. Gliwice - Poland; ³Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona, Verona - Italy;

⁴CRIAMS-Sport Medicine Centre Voghera University of Pavia, Voghera - Italy

PLENARY ROOM

18:30 - 20:00 **General Assembly**

20:30 - 23:00 **Social Dinner**



Saturday, May 31, 2025

PLENARY ROOM

08:00 - 09:00 GDS Meetings

Myotonic Dystrophies and Myotonias | **PLENARY ROOM**
 Rehabilitation and Long-Term Care | **PARALLEL ROOM 1**
 Laminopathies Group | **PARALLEL ROOM 2**

09:00 - 09:30 **Lecture 3**

Chairperson: *Stefania Corti (Milano)*
 In vitro platforms to study neuromuscular diseases:
 challenges and opportunities
Francesco Saverio Tedesco (Londra)

PARALLEL ROOM 1

09:00 - 09:30 **Lecture 4**

Chairperson: *Massimo Domenico Zeviani (Padova)*
 Updates on clinical trials in Mitochondrial Myopathies
Thomas Klopstock (Monaco)

PLENARY ROOM

09:30 - 10:30 **ORAL COMMUNICATION | SPINAL MUSCLE ATROPHY (SMA)**
 Chairpersons: *Caterina Agosto (Padova), Enrico Bertini (Roma)*

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

09:30 - 09:42 **TIME-DEPENDENT SMN2 SPLICING MODULATION RESTORES MOTOR NEURON FUNCTION IN SMA SPINAL CORD ORGANOIDs THROUGH CILIARY GENE REGULATION**

Corti S.¹, D'Angelo A.¹, Beatrice F.¹, Cordiglieri C.², Miotto M.³, Lodato S.³, Rizzo F.¹, Comi G.¹, Ottoboni L.¹, Parente V.⁴

¹University of Milan, Milano - Italy; ²INGM, Milan - Italy; ³Humanitas University, Milano - Italy; ⁴Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico

09:42 - 09:54 **DYSPHAGIA IN SMA1: CLINICAL FEATURES AND ASSESSMENT TOOLS**

Gandolfi S.¹, Dosi C.¹, Parravicini S.¹, Scopelliti M.R.¹, Zanin R.¹, Arnoldi M.T.¹, Schilder A.², Masson R.¹

¹Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milano - Italy; ²Department of Biomedical and Clinical Sciences, Università degli Studi di Milano, Milan - Italy

09:54 - 10:06 **HAS RESPIRATORY FUNCTION ASSESSMENT HIGHER RESPONSIVENESS THAN MOTOR FUNCTIONAL EVALUATION TO THE POSITIVE EFFECT OF RISDIPLAM IN LATER-ONSET SMA PATIENTS? THE EXPERIENCE OF A LONG-TERM FOLLOW-UP IN TWO ITALIAN CENTERS**

Sframeli M.¹, Brolatti N.², Trucco F.³, La Rosa E.¹, Lauro R.¹, Matera R.¹, Maniaci M.¹, Longoni N.¹, Said I.¹, Allegra C.¹, Pedemonte M.³, Bruno C.², Messina S.¹

¹Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy;
²Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova - Italy; ³Paediatric Neurology and Muscular Diseases Unit, IRCCS Istituto Giannina Gaslini and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Genova - Italy

10:06 - 10:18 LONELINESS AND QUALITY OF LIFE IN ADULT SMA PATIENTS: THE EXPERIENCE OF THE PARMA CENTRE

Libelli G.¹, Nuredini A.¹, D'Orsi A.¹, Allegri I.², Chierici E.², Saccani E.²

¹Unit of Neurology, Department of Medicine and Surgery, University of Parma, Parma, Italy;

²Neurology Unit, Department of General and Specialized Medicine, University Hospital of Parma, Parma - Italy

10:18 - 10:30 COULD ORSAT HELP TO IMPROVE THE MULTIDIMENSIONAL CARE IN SPINAL MUSCULAR ATROPHY ADVANCED DISEASE STAGE?

Maniaci M.¹, Longoni N.¹, Materia R.¹, La Rosa E.¹, Lauro R.¹, Ibrahim S.¹, Allegra C.¹, Sframeli M.¹, Mercuri E.², Messina S.¹

¹Department of Clinical and Experimental Medicine, University of Messina, Messina - Italy ;

²Child Neurology and Psychiatry, Catholic University of Rome, Roma - Italy

PARALLEL ROOM 1

09:30 - 10:30 ORAL COMMUNICATION | MYOTONIC DYSTOPHIES

Chairpersons: Giovanni Meola (Milano), Roberto Massa (Roma)

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

09:30 - 09:42 EXCESSIVE DAYTIME SLEEPINESS IN MYOTONIC DYSTROPHY TYPE 1 (DM 1): CAN MELATONIN PLAY A ROLE?

Colacicco G., Ferrari Aggradi C., Zanolini A., Lazzaro C., Di Natale E., Roma E., Sansone V.A.

The NeMO Center- Milan, Neurorehabilitation Unit University of Milan Milan Italy

09:42 - 09:54 SETTING THE BASIS FOR A NATIONAL MYOTONIC DYSTROPHY REGISTRY: THE NEMO EXPERIENCE TOWARDS TRIAL READINESS

Ferrari Aggradi C.R.¹, Coccia M.², Filosto M.³, Limongelli G.⁹, Mercuri E.⁴, Pane M.⁴, Patanella A.K.⁴, Conte A.⁴, Sabatelli M.⁴, Zuccarino R.⁵, Barp A.⁵, Rao F.⁶, Liguori R.⁷, Zanolini A.⁸, Colacicco G.⁸, Sansone V.A.⁸

¹Università degli Studi di Milano, Milano - Italy; ²Centro Clinico Nemo Ancona, Ancona

- Italy; ³Centro Clinico Nemo Brescia, Brescia - Italy; ⁴Centro Clinico NeMO Roma, Roma

- Italy; ⁵Centro Clinico NeMO Trento, Trento - Italy; ⁶Centro Clinico NeMO Arenzano,

Arenzano - Oman; ⁷Centro Clinico NeMO Bologna, Bologna - Italy; ⁸Centro Clinico NeMO

Milano, Milano - Ital; ⁹Centro Clinico NeMO Napoli, Napoli - Italy

09:54 - 10:06 COMPUTER ASSISTED COGNITIVE TESTS: THE ROLE OF COGSTATE BRIEF BATTERY IN MYOTONIC DYSTROPHY TYPE 1

Sansone V.A.¹, Ferrari Aggradi C.¹, Greco L.¹, Beretta M.¹, Pozzi S.¹, Casiraghi J.¹, Zanolini A.¹, Colacicco G.¹, Hung M.², Berggren K.², Ikegami K.², Johnson N.²

¹The NeMO Clinical Center in Milan, Neurorehabilitation Unit, University of Milan, Milan -

Italy; ²Virginia Commonwealth University, Neuromuscular Unit, Department of Neurology,

Virginia, USA, Richmond - United States of America

10:06 - 10:18 TEST-RETEST RELIABILITY OF REMOTE ASSESSMENTS IN MYOTONIC DYSTROPHY TYPE 1

Tufano L.¹, Richardson E.², Dekdebrun J.², Stanton E.², Docteur M.², Eichinger K.², Thornton C.², Hamel J.²

¹Sapienza University of Rome, Neuromuscular and Rare Disease Centre, Neurology, Sant'Andrea Hospital, Rome- Italy; ²University of Rochester Medical Center, Neurology Rochester, NY - United States of America

10:18 - 10:30 EFFICACY AND SAFETY OF METFORMIN IN MYOTONIC DYSTROPHY TYPE 1 (DM1). THE METMYD STUDY FINAL RESULTS

Frezza E.¹, Rossi S.², Perna A.³, Bucci E.⁴, Greco G.¹, Goglia M.¹, Tufano L.⁴, Riso V.², Visconti V.V.⁵, Botta A.⁵, Nuccetelli M.⁶, Antonini G.⁷, Petrucchi A.³, Silvestri G.², Massa R.¹

¹Unit of Neuromuscular Diseases, Department of Systems Medicine, Tor Vergata University of Rome, Rome - Italy; ²Institute of Neurology, Department of Neuroscience, Catholic University of the Sacred Heart, Rome - Italy; ³Center for Neuromuscular and Neurological Rare Diseases, San Camillo-Forlanini Hospital, Rome - Italy; ⁴Unit of Neuromuscular Diseases, Sant'Andrea Hospital, Rome - Italy; ⁵Unit of Medical Genetics, Department of Biomedicine and Prevention, Tor Vergata University of Rome, Rome - Italy; ⁶Department of Laboratory Medicine, Tor Vergata University Hospital, Rome - Italy; ⁷Unit of Neuromuscular Diseases, Department of Neurology Mental Health and Sensory Organs (NESMOS), Faculty of Medicine and Psychology, 'Sapienza' University of Rome, Sant'Andrea Hospital, Rome - Italy

PARALLEL ROOM 2**09:30 - 10:30 ORAL COMMUNICATION: MYOSITIS AND OTHERS**

Chairpersons: *Chiara Fiorillo (Genova), Marcello Romano (Palermo)*

Each Presenting Author will be given 10 minutes for presentation and 2 minutes for discussion

09:30 - 09:42 QUANTITATIVE MUSCLE ULTRASOUND IN INCLUSION BODY MYOSITIS

Costanzo R.¹, Falco P.¹, Garibaldi M.², Lauletta A.², Truini A.¹, Evangelisti E.¹, Di Pietro G.¹

¹Department of Human Neuroscience - Sapienza University, Rome - Italy; ²Neuromuscular and Rare Disease Centre, Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University, Sant'Andrea Hospital, Rome - Italy

09:42 - 09:54 EVIDENCE FOR A CONTRIBUTION OF EOTAXIN FAMILY MEMBERS TO INCLUSION BODY MYOSITIS: NOVEL DISEASE BIOMARKERS AND IMMUNOPATHOGENETIC MEDIATORS

Riolo G.¹, Salerno F.¹, Vicino A.², Cheli M.¹, Gibertini S.¹, Ruggieri A.¹, Iannibelli E.¹, Carnazzi A.¹, Nicolini De Gaetano L.¹, Bonanno S.¹, Marcuzzo S.¹, Cavalcante P.¹, Maggi L.¹

¹Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan - Italy; ²Nerve-Muscle Unit - Neurology Service, Department of Clinical Neurosciences, Lausanne University Hospital and University of Lausanne, Lausanne - Swaziland

09:54 - 10:06 NEUROMUSCULAR ASSEMBLOIDS AS PLATFORMS TO STUDY MUSCULAR DYSTROPHIES

Fontanelli L.¹, Vadi G.², Ricci G.², Siciliano G.²

¹Health Science Interdisciplinary Center, Sant'Anna School of Advanced Studies, Pisa - Italy;

²Department of Clinical and Experimental Medicine, University of Pisa, Pisa - Italy

10:06 - 10:18 EULAR/ACR CORE SET MEASURES IN IIM: WHEN ONE SIZE DOES NOT FIT ALL - THE CASES OF THE ILIOPSOAS AND ASYMPTOMATIC MYOCARDITIS

Laurini C.¹, Bosco L.¹, Falzone Y.M.¹, Ratti A.¹, Napolitano A.², Canti V.³, Fumagalli A.³, Del Carro U.⁴, Filippi M.¹, Gerevini S.², Rovere Querini P.³, Previtali S.C.¹

¹Department of Neurology, IRCCS San Raffaele Scientific Institute Milan Italy, ²Department of Neuroradiology, ASST Papa Giovanni XXIII Bergamo Italy, ³San Raffaele Scientific Institute, Division of Immunology, Transplantation, and Infectious Diseases Milan Italy,

⁴Neurophysiology Service, IRCCS San Raffaele Scientific Institute Milan Italy

10:18 - 10:30 UNDER THE LENS: A SINGLE-CENTER RETROSPECTIVE ANALYSIS OF MUSCLE BIOPSIES IN THE LAST DECADE

Urbano G.¹, Vercelli L.¹, Gadaleta G.¹, Boschi S.¹, Leopizzi E.¹, D'Alessandro R.², Ricci F.S.², Mongini T.E.¹

¹Neuromuscular Unit, Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin - Italy; ²Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, University of Turin, Turin - Italy

10:30 - 11:00 Coffee Break

PLENARY ROOM

11:00 - 12:00 MUSCLE TRIVIAM CONTEST | FINAL

Chairpersons: *Claudio Bruno (Genova), Serenella Servidei (Roma), Antonio Trabacca (Brindisi)*



**12:00 - 13:30 ROUND TABLE MEETING with PATIENT ASSOCIATIONS
MALATTIE NEUROMUSCOLARI:
BISOGNI, ASPETTATIVE E PROSPETTIVE DI CURA**

Chairpersons: *Federica Silvia Ricci (Torino), Vincenzo Nigro (Napoli), Luisa Politano (Napoli)*



*Elisabetta Conti
Francesco Ieva
Renato De Regibusper
Anita Pallara
Danila Baldessari
Beatrice Vola
Fabrizio Farnetani
Fernanda De Angelis
Stefania Pedroni*

**AIG
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Consulta Malattie Neuromuscolari ETS
Famiglia SMA
Fondazione Telethon
GFB Gruppo Familiari Beta-sarcoglicanopatie
Mitocon
Parent Project
UILDM**

13:30 Closing Ceremony & Awards

XXV AIM National Congress

POSTER SESSION

Poster Session | NO CME

Each Presenting Author will be given 5 minutes for presentation and 2 minutes for discussion

Spinal Muscular Atrophies and Moto-Neuron Disorders

P01 - P19

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: Stefano Previtali (Milano), Federica Trucco (Genova)

P01 - NEUROMUSCULAR TAPING FOR DROP HAND REHABILITATION IN AN EARLY-TREATED INFANT WITH SMA1: A CASE REPORT

Branca A.^[1], Saraceni V.^[1], Pelliccioni M.^[1], Onofri M.^[1], Sancricca C.^{[1][2]}, Frongia A.L.^{[1][3]}

[1] Fondazione UILDM Lazio Onlus - Rome - Italy, [2] Neurophysiopathology unit, Fondazione Policlinico Universitario A. Gemelli IRCCS - Rome - Italy, [3] Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy

P02 - SUNFISH PARTS 1 AND 2: 5-YEAR SAFETY AND EXPLORATORY EFFICACY DATA OF RISDIPLAM IN TYPES 2 AND 3 SPINAL MUSCULAR ATROPHY

Masson R.^[1], Servais L.^[2], W Day J.^[3], Deconinck N.^[4], Mazzone E.S.^[5], Nascimento A.^[6], Oskoui M.^[7], Saito K.^[8], Vuillerot C.^[9], Baranello G.^[10], Boespflug-Tanguy O.^[11], Goemans N.^[12], Kirschner J.^[13], Kostera-Pruszyk A.^[14], Sully K.^[15], Kuthiala M.^[16], Gorni K.^[17], Martin C.^[15], Yin Yeung W.^[15], Scalco R.S.^[16], Mercuri E.^[5]

[1] Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy, [2] MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford, Oxford, UK - Oxford - United Kingdom, [3] Department of Neurology, Stanford University, Palo Alto, CA, USA - Palo Alto - United States of America, [4] Neuromuscular Reference Center, UZ Gent, Ghent, Belgium - Ghent - Belgium, [5] Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, Rome, Italy; - Roma - Italy, [6] Neuromuscular Unit, Neuropaediatrics Department, Hospital Sant Joan de Déu, Fundacion Sant Joan de Déu, CIBERER - ISC III, Barcelona, Spain - Barcellona - Spain, [7] Departments of Pediatrics and Neurology Neurosurgery, McGill University, Montreal, Canada; - Montreal - Canada, [8] Institute of Medical Genetics, Tokyo Women's Medical University, Tokyo, Japan - tokyo - Japan, [9] Service de Rééducation Pédiatrique Infantile "L'Escale", Hôpital Mère Enfant, CHU-Lyon, Bron, France - Lyon - France, [10] The Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health University College London & Great Ormond Street Hospital Trust, London, UK - London - United Kingdom, [11] I-Motion, Institut de Myologie AP-HP, Hôpital Armand Trousseau, Paris, France - Paris - France, [12] Neuromuscular Reference Centre, Department of Paediatrics and Child Neurology, University Hospitals Leuven, Leuven, Belgium - Leuven - Belgium, [13] Department of Neuropediatrics and Muscle Disorders, Faculty of Medicine, Medical Center-University of Freiburg, Freiburg, Germany - Freiburg - Germany, [14] Department of Neurology, Medical University of Warsaw and ERN EURO-NMD, Warsaw, Poland - Warsaw - Poland, [15] Roche Products Ltd, Welwyn Garden City, UK - Welwyn - United Kingdom, [16] Pharma Development, Safety, F. Hoffmann-La Roche Ltd, Basel, Switzerland - Basel - Switzerland, [17] PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland - Basel - Switzerland

P03 - RAINBOWFISH: PRIMARY EFFICACY AND SAFETY DATA IN RISDIPLAM-TREATED INFANTS WITH PRESYMPTOMATIC SPINAL MUSCULAR ATROPHY (SMA)

Bertini E.^{*(1)}, Finkel R.S.^[2], Laurent S.^[3], Vlodavets D.^[4], Edmar Z.^[5], Al--Muhaizea M.^[6], Araújo A.P.^[7], Nelson L.^[8], Jaber B.^[9], Gorni K.^[10], Kletzl H.^[11], Palfreeman L.^[12], Gaki E.^[12], Rabbia M.^[13], Summers D.^[12], Fontoura P.^[10], Farrar M.A.^[14]

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Centre, Department of Paediatrics, University of Oxford, Oxford, UK - Oxford - United Kingdom, [4]Russian Children Neuromuscular Center, Veltischev Clinical Pediatric Research Institute of Pirogov Russian National Research Medical University, Moscow, Russia; - Moscow - Russian Federation, [5]Department of Neurology, Faculdade de Medicina, Universidade de São Paulo, São Paulo, Brazil - Sao Paulo - Brazil, [6]Department of Neurosciences, King Faisal Specialist Hospital & Research Center-Riyadh, Riyadh, Kingdom of Saudi Arabia - Riyadh - Saudi Arabia, [7]Pediatrics Department, Faculty of Medicine, Federal University of Rio de Janeiro, Rio de Janeiro, Brazil - Rio de Janeiro - Brazil, [8]Department of Physical Therapy, University of Texas Southwestern Medical Center, Dallas, TX, USA; - Dallas - United States of America, [9]Pharma Development, Safety, F. Hoffmann-La Roche Ltd, Basel, Switzerland - Basel - Switzerland, [10]PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland - Basel - Switzerland, [11]Roche Pharmaceutical Research and Early Development, Roche Innovation Center Basel, Basel, Switzerland - Basel - Switzerland, [12]Roche Products Ltd, Welwyn Garden City, UK - Welwyn - United Kingdom, [13]Genentech, Inc., South San Francisco, CA, USA - San Francisco - United States of America, [14]Sydney Children's Hospital Network and UNSW Medicine, UNSW Sydney, Sydney, Australia - Sydney - Australia

P04 - A NEW REHABILITATION APPROACH FOR ADULT SMA PATIENTS: THE ROLE OF NEUROPLASTICITY

De Maria M., Zoppi D., Russo A., Vallefucio F., Iodice R., Esposito G., Manganelli F., Ruggiero L.
Department of Neurosciences, Reproductive Sciences, and Odontostomatology, University of Naples
"Federico II," Naples, Italy - Napoli - Italy

P05 - FROM CARE TO FOLLOW-UP OF THE CHILD WITH SPINAL MUSCULAR ATROPHY (SMA): SHARED NURSING PATH

Bruscino G.^[1], Bravetti C.^[2], Carta C.^[3], Cosma L.^[4], Cellamare P.^[5], Fontana M.^[6], Genzone P.^[7], Gotti E.^[8], Mangia E.D.^[9], Palumbo N.^[5], Pellegrino M.^[10], Rufini E.^[11], Sarracino F.^[1], Santalucia P.^[12]

[1]Pediatric Neurology, Santobono-Pausilipon Children's Hospital - Napoli - Italy, [2]Pediatric Neurology, IRCCS Agostino Gemelli University Hospital - Roma - Italy, [3]Pediatric Neurology, University Hospital of Padua - Padova - Italy, [4]Pediatric Oncohematology Unit, University Hospital of Padua - Padova - Italy, [5]Pediatric Neurology, Giovanni XXIII Children's Hospital - Bari - Italy, [6]DH Pediatric Neurology and Neuromuscular Diseases, IRCCS Giannina Gaslini Institute - Genova - Italy, [7]DH Muscle and Neurodegenerative Diseases, Bambino Gesù Children's Hospital, IRCCS - Roma - Italy, [8]Fondazione Serena Onlus, Neuromuscular Omni Centre (NeMo) - Milano - Italy, [9]IRCCS Neurological Institute Carlo Besta - Milano - Italy, [10]Child Neuropsychiatry Unit, Regina Margherita Children's Hospital - Torino - Italy, [11]Pneumology and Cystic Fibrosis Unit, Bambino Gesù Children's Hospital, IRCCS - Roma - Italy, [12]San Giovanni Addolorata Hospital - Roma - Italy

P06 - COGNITIVE FUNCTION IN SMA2 AND SMA3 PATIENTS: INVESTIGATING ATTENTION AND SOCIAL PERCEPTION WITH NEPSY-II

Khalil S.^[1], Iosca A.^[1], Carraro L.^[1], Conti C.^[1], Gardani A.^[2], Berardinelli A.^[2]

[1]Department of Brain and Behavioral Sciences, University of Pavia - Pavia - Italy, [2]Department of Child Neurology and Psychiatry, IRCCS C. Mondino Foundation - Pavia - Italy

P07 - VIRTUAL REALITY AND TELE-REHABILITATION IN NEUROMOTOR REHABILITATION: CASE STUDIES OF SPINAL MUSCULAR ATROPHY TYPES 2 AND 3 DURING GENE-BASED THERAPY

Pirani G.^[1], Accogli G.^[2], Macchitella L.^[1], Barraco G.^[1], Ruggiero M.^[1], Giannotta G.^[1], Oliva M.C.^[1], Ferrante C.^[1], Trabacca A.^[1]

[1]Scientific Hospital for Neurorehabilitation - Unit for Severe Disabilities in Developmental Age and Young Adults (Developmental Neurology and Neurorehabilitation) - Associazione "La Nostra Famiglia" - IRCCS "E. Medea" - 72100 Brindisi, Italy - Brindisi -, [2]Scientific Institute IRCCS "E. Medea", Scientific Direction, 23842 Bosisio Parini, Italy - Brindisi - Italy

P08 - OPTIMIZING MULTIDISCIPLINARY CARE FOR SPINAL MUSCULAR ATROPHY: A PILOT STUDY ON GOAL-SETTING USING THE INTERNATIONAL CLASSIFICATION OF FUNCTIONING, DISABILITY, AND HEALTH

Ruggiero M., Giannotta G., Pirani G., Oliva M.C., Ferrante C., Trabacca A.

Scientific Hospital for Neurorehabilitation - Unit for Severe Disabilities in Developmental Age and Young Adults (Developmental Neurology and Neurorehabilitation) - Associazione "La Nostra Famiglia" - IRCCS "E. Medea" - 72100 Brindisi, Italy - Brindisi -

P09 - MYOSTATIN LEVELS IN SMA FOLLOWING DISEASE-MODIFYING TREATMENTS: A MULTI-CENTER STUDY

Capasso A.^[1], Piemonte F.^[2], Petrillo S.^[2], Coratti G.^[1], D'Amico A.^[2], Catteruccia M.^[2], Pera M.C.^[1], Palermo C.^[1], Pane M.^[1], Abiusi E.^[3], Cicala G.^[1], Villa M.^[1], Bravetti C.^[1], Arpaia C.^[1], Novelli A.^[3], Fanqui S.^[3], Fiori S.^[3], Napoli G.^[4], Baroni S.^[4], Tiziano F.D.^[3], Bertini E.S.^[2], Comi G.^[5], Corti S.^[5], Mercuri E.^[1]

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P10 - TRIO RNA-SEQUENCING IN CEREBROSPINAL FLUID OF NUSINERSEN-TREATED ADULT SMA PATIENTS: A GLIMPSE INTO TITIN AND NEBULIN DEREGULATION

Dragoni F., Rizzo B., Dainesi M.I., Di Gerlando R., Minucchi E., Asteggiano C., Pichiecchio A., Berardinelli A.L., Gagliardi S.

IRCCS Mondino Foundation - Pavia - Italy

P11 - MINIMALLY INVASIVE FUSIONLESS SURGERY FOR SCOLIOSIS IN SPINAL MUSCULAR ATROPHY

Catteruccia M.^[1], Iorio C.^[2], Tosi M.^[1], De Luca G.^[1], Mizzone I.^[1], Carlesi A.^[3], Bertini E.^[1], Mazza O.^[2], D'Amico A.^[1]

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P12 - SCOLIOSIS IN SMA PATIENTS TREATED WITH DISEASE-MODIFYING DRUGS: A PRELIMINARY STUDY

Ait Allali N.^[1], Risi B.^[1], Damioli S.^[1], Garletti G.^[1], Giovanelli G.^[1], Colombi C.^[1], Caria F.^[1], Garofali F.^[1], Ottelli E.^[1], Carugati R.^[1], Poli L.^[2], Padovani A.^[3], Filosto M.^[1]

[1]NeMO-Brescia Clinical Center for Neuromuscular Diseases - Brescia - Italy, [2]Unit of Neurology, ERN EURO-NMD Center ASST Spedali Civili - Brescia - Italy, [3]Department of Clinical and Experimental Sciences, University of Brescia - Brescia - Italy

P13 - RISDIPLAM IMPACT ON RESPIRATORY FUNCTION IN PATIENTS WITH SPINAL MUSCULAR ATROPHY TYPES 2 AND 3: A 1-YEAR RETROSPECTIVE BICENTRIC STUDY

Falco A.^[1], Malaspina M.^[1], Roller E.^[1], Venditti R.^[1], Coratti G.^[2], Paolucci S.^[2], Cutri C.^[2], Franchino V.^[3], Giacompolli I.^[3], Lazzaro C.^[3], Albamonte E.^[3], Sansone V.^[3], Pane M.^[2], Mercuri E.M.^[2]

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P14 - RISDIPLAM THERAPY: AN OBSERVATIONAL FOUR-YEAR STUDY IN A SMA PAEDIATRIC COHORT

Tosi M.^[1], Catteruccia M.^[1], Diodato D.^[1], Carlesi A.^[2], De Luca G.^[1], Mizzoni I.^[1], Longo A.^[1], Bertini E.S.^[1], D'Amico A.^[1]

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[2]Unit of Developmental Neurology, IRCCS Bambino Gesù Children Hospital - Rome - Italy

P15 - CLINICAL AND NEUROPHYSIOLOGICAL FOLLOW-UP OF SPINAL MUSCULAR ATROPHY PATIENTS TREATED WITH RISDIPLAM: A REAL-LIFE SINGLE CENTER STUDY

Rini N., Alonge P., Lupica A., Crescimanno G., Di Stefano V., Torrente A., Brighina F.

Department of Biomedicine, Neuroscience, and advanced Diagnostic (BIND), University of Palermo - Palermo

- Italy

P16 - FROM MOVEMENT TO AUTONOMY: EVALUATING CHANGES IN ADL AND IADL IN SMA PATIENTS TREATED WITH DISEASE-MODIFYING THERAPIES

Carraro L.^[1], Iosca A.^[1], Dainesi M.I.^[1], Khalil S.^[1], Conti C.^[1], Gardani A.^[2], Berardinelli A.^[2]

[1]Department of Neuroscience and Behavioral Sciences, University of Pavia - Pavia - Italy, [2]Child

Neuropsychiatry, IRCCS C. Mondino Foundation - Pavia - Italy

P17 - TWO YEARS OF RISDIPLAM TREATMENT IN ADULTS: INSIGHTS FROM A SINGLE-CENTER EXPERIENCE

Alonge P., Lupica A., Rini N., Brighina F.

Department of Biomedicine, Neuroscience and Advanced Diagnostic (BIND), University of Palermo, - Palermo

- Italy

P18 - IMPACT OF DISEASE-MODIFYING TREATMENT ON RESPIRATORY OUTCOMES AFTER SCOLIOSIS SURGERY IN SMA PAEDIATRIC PATIENTS

Morando S.^[1], Trucco F.^[2], Brolatti N.^[1], Panicucci C.^[1], Latinakis G.^[1], Pedemonte M.^[3], Bruno C.^[4]

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[2]Paediatric Neurology and Muscle Diseases Unit, IRCCS Istituto Giannina Gaslini, Genoa and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova - Genova - Italy, [3]Pediatric Neurology and Muscle Diseases Unit, IRCCS Istituto Giannina Gaslini, Genova - Genova - Italy, [4]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova

and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova - Genova - Italy

P19 - AN ITALIAN CASE OF SPINAL MUSCULAR ATROPHY WITH LOWER EXTREMITY DOMINANCE 2A DUE TO A DE NOVO MUTATION PRESENTING A FEW UNUSUAL FEATURES.

Barbaccia A.^[1], Pugliese A.^[1], Biasini F.^[1], Tortorella A.^[2], Nigro V.^[2], Musumeci O.^[1], Rodolico C.^[1]

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Metabolic Myopathies - Mitochondrials P20 - P35

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: Ignazio Arena (Messina), Daria Diodato (Roma)

P20 - AN INHERITED MTDNA REARRANGEMENT, MIMICKING A SINGLE LARGE-SCALE DELETION, ASSOCIATED WITH MIDD AND A PRIMARY CARDIOLOGICAL PHENOTYPE

Lopriore P.^[1], Legati A.^[2], Neuhofer C.M.^[3], Lo Gerfo A.^[4], Kopajtich R.^[5], Barresi M.^[2], Cecchi G.^[1], Pavlov M.^[5], Izzo R.^[2], Montano V.^[1], Caligo M.A.^[4], Berutti R.^[5], Mancuso M.^[1], Prokisch H.^[5], Ghezzi D.^[2]

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P21 - AUTOPHAGY IMPAIRMENT CORRELATES WITH ENHANCED SATELLITE CELL ACTIVATION IN MUSCLE BIOPSIES FROM YOUNGER LATE ONSET POMPE DISEASE (LOPD) PATIENTS

Severa G.^{[1][2]}, Bastu S.^[1], Borin G.U.^{[1][3]}, Decrouy X.^[1], Codina A.^[4], Kefi K.^[1], Periou B.^{[1][2]}, Nadaj-Pakleza A.^[5], Lannes B.^[5], Sacconi S.^[6], Maurage C.^[7], Tard C.^[5], Jou C.^[7], Nasciminto Osorio A.^[4], Taglietti V.^[1], Malfatti E.^{[1][2]}

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P22 - MOBILE HEALTH TECHNOLOGY IN LATE-ONSET POMPE DISEASE: SHORT-TERM FOLLOW-UP AFTER SWITCHING TO AVALGLUCOSIDASE ALPHA

Risi B.^[1], Ferullo L.^[2], Olivieri E.^[2], Labella B.^[2], Rizzardi A.^[3], Caria F.^[4], Ait Allali N.^[4], Franchi E.^[4], Poli L.^[5], Bozzoni V.^[5], Padovani A.^[2], Pilotto A.^[6], Filosto M.^[7]

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P23 - THE 9 PB DELETION BETWEEN THE MITOCHONDRIAL COII AND LYSINE TRNA GENES IN CAUCASIAN POPULATION WITH COGNITIVE DISORDERS

Lo Giudice M., Giuliano M., Santa Paola S., Borgione E., Di Blasi F.D., Pettinato R., Romano C., Scuderi C.

IRCCS Oasi Maria SS - Troina - Italy

P24 - EARLY ONSET OF NEUTRAL LIPID STORAGE DISEASE WITH MYOPATHY IN A HUNGARIAN FAMILY.

Angelini C.^[1], Martegani E.^[2], Missaglia S.^[2], Fylymonenko V.^[2], Taviani D.^[2]

[1]University of Padova - Padova - Italy, [2]Catholic University - Milan - Italy

P25 - RAPAMYCIN TREATMENT IN A MERRF PATIENT

La Morgia C.^[1], Manzoni E.^[1], Moresco M.^[2], Maresca A.^[2], Capristo M.A.^[2], Romagnoli M.^[2], Carelli V.^[1]

[1]University of Bologna - Bologna - Italy, [2]Programma di Neurogenetica, IRCCS Istituto delle Scienze Neurologiche di Bologna - Bologna - Italy

P26 - CLINICALLY IMPORTANT IMPROVEMENTS IN 6-MINUTE WALK DISTANCE AND FORCED VITAL CAPACITY IN ADULTS WITH LATE-ONSET POMPE DISEASE SWITCHING FROM ALGLUCOSIDASE ALFA TO CIPAGLUCOSIDASE ALFA PLUS MIGLUSTAT IN THE PROPEL STUDY

Vagni F.^[1], Kushlaf H.^[2], Schoser B.^[3], Wenninger S.^[3], Hummel N.^[4], Clarke S.^[5], Crabtree M.^[5], Fox B.^[6], Gloeckner B.^[7], Holdbrook F.^[6], Jain V.^[6], Peceny M.^[7], Claeys K.G.^[8]

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P27 - SUDDEN ONSET OF CYCLIC DIARRHEA IN POLG1 NEUROMYOPATHY: A TREATMENT CHALLENGE.

Diodato D.^[1], Angelino G.^[2], Tambucci R.^[2], Chiarini B.^[3], Tosi M.^[1], Catteruccia M.^[1], De Angelis P.^[2], Bertini E.S.^[1], D'Amico A.^[1]

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P28 - EARLY EFFECTIVENESS AND SAFETY OF OMAVELOXOLONE IN FRIEDREICH'S ATAXIA: A REAL-WORLD STUDY FROM PALERMO

Lima S.M., Caltagirone M., Rini N., Quartetti U., D'Amico F., Messina C., D'Alonge P., Brighina F., Di Stefano V.

Department of Biomedicine, Neuroscience, and Advanced Diagnostic (BIND), University of Palermo, Palermo, Italy. - Palermo - Italy

P29 - VERY LATE-ONSET LIPID STORAGE MYOPATHY DUE TO LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (LCHADD)

Melilli C.^[1], Porcino M.^[1], Licitri L.^[1], Usbergo C.^[1], Bannino M.C.^[1], Rodolico C.^[1], Santorelli F.M.^[2], Musumeci O.^[1]

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P30 - NEURODEGENERATIVE AND INFLAMMATORY BIOMARKERS IN MITOCHONDRIAL DISEASES: INSIGHTS INTO DISEASE MECHANISMS AND SEVERITY

Arena I.G., Harrison E., Ratnaïke T., Schon K., Olimpio C., Golder Z., Ashby E., Addy C., Biggs H., Van Den Ameele J., Yu Wai Man P., Horvath R., Chinnery P.

Department of Clinical Neurosciences, University of Cambridge, Cambridge, UK - Cambridge - United Kingdom

P31 - MULTIMODAL EVALUATION OF UNRELATED MYOPATHY CASES CARRYING A SINGLE MUTATION IN THE ALPHA-GLUCOSIDASE GENE: A CASE SERIES.

Russo A., Zoppi D., Vallefucio F., De Maria M., Russo R., Boemia V., Iapoce M., Vitale F., Ruggiero L.

Department of Neurosciences, Reproductive Sciences, and Odontostomatology, University of Naples "Federico II". - Napoli - Italy

P32 - ANALYSIS OF GAIT SPATIAL-TEMPORAL PARAMETERS IN PATIENTS AFFECTED BY LATE-ONSET POMPE DISEASE (LOPD)

Solla P.^[1], Ercoli T.^[2], Leoni S.^[2], Ruiu E.^[2], Castiglia F.S.^[3], Occhipinti C.^[4], Della Croce U.^[4], Picerno P.^[4]

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P33 - MITOCHONDRIAL HOMEOSTASIS AND QUALITY CONTROL IN PERIPHERAL NEUROPATHIES: EXPLORING THE MTOR/AKT AXIS IN CMT2A2, ADOA, AND CMT4B3

Zanfardino P.^[1], Amati A.^[1], Santorelli F.M.^[2], Petruzzella V.^[1]

[1]Dipartimento di Biomedicina traslazionale e Neuroscienze Università degli studi di Bari Aldo Moro - Bari - Italy, [2]U.O.C. Medicina Molecolare, Neurogenetica e Malattie Neuromuscolari IRCCS Fondazione Stella Maris

P34 - RAPIDLY PROGRESSIVE SENSORY NEUROPATHY AND PANCYTOPENIA REVEALING ATYPICAL ADULT-ONSET MULTIPLE ACYL-COENZYME A DEHYDROGENASE DEFICIENCY (MADD)

Forcina F., Marando D., Leonardi L., Fionda L., Merlonghi G., Costanzo R., Vera V., Lauletta A., Rossini E., Tufano L., Morino S., Antonini G., Garibaldi M.

Sapienza University of Rome - Rome - Italy

P35 - SERTRALINE-ASSOCIATED MULTIPLE ACYL-COA DEHYDROGENASE DEFICIENCY (MADD): A NOVEL NOSOLOGICAL ENTITY

Marando D.^[1], Lauletta A.^[1], Merlonghi G.^[1], Fionda L.^[2], Leonardi L.^[2], Morino S.^[2], Rossini E.^[1], Forcina F.^[1], Antonini G.^[1], Garibaldi M.^[1]

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Myasthenia Gravis I P36 - P54



Friday, May 30, 2025 15:45 - 17:30

Chairpersons: *Filippo Brighina (Palermo), Elena Pennisi (Roma)*

P36 - REAL WORLD STUDY IN ITALIAN PUBLIC HOSPITAL WITH EFGARTIGIMOD IN PATIENTS AFFECTED BY GENERALIZED MYASTHENIA GRAVIS.

Sgarzi M., Paone P., Camera G., Agazzi E., Alimonti D.

ASST PAPA GIOVANNI XXIII - Bergamo - Italy

P37 - COMPARISON OF CLINICAL RESPONSE WITH MG-ADL TO INNOVATIVE BIOLOGICAL THERAPIES IN GENERALIZED MYASTHENIA GRAVIS

Messina C., Rini N., Alonge P., Luppino P., Bonaventura A., Di Martino G., Campo S., Brighina F., Di Stefano V.

Department of Biomedicine, Neuroscience and Advanced Diagnostics (Bi.N.D.), University of Palermo, 90127 Palermo, PA, Italy. - Palermo - Italy

P38 - RESPONSE INFLAMMATORY INDEXES AS PREDICTIVE MARKERS FOR CLINICAL RESPONSE TO INNOVATIVE BIOLOGICAL THERAPIES IN GENERALIZED MYASTHENIA GRAVIS

Messina C., Rini N., Alonge P., Luppino P., Bonaventura A., Di Martino G., Campo S., Brighina F., Di Stefano V.

Department of Biomedicine, Neuroscience and Advanced Diagnostics (Bi.N.D.), University of Palermo, 90127 Palermo, PA, Italy. - Palermo - Italy

P39 - REAL-WORLD REDUCTION IN ORAL CORTICOSTEROID UTILIZATION AT 1-YEAR FOLLOWING EFGARTIGIMOD INITIATION

Goyal N.^[1], De Notariis A.M.^[2], Qi C.^[2], Stone J.^[3], Brauer E.^[2], Jefferson M.^[2], Balaji Suthagar T.^[4], Menon R.R.^[4], Sato M.^[5], Phillips G.^[2]

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P40 - EFGARTIGIMOD IN GENERALIZED MYASTHENIA GRAVIS. REAL-LIFE EXPERIENCE FROM A SINGLE CENTER.

D'Amico F., Campo S., Bonaventura A., Rini N., Alonge P., Portera V., Quartetti U., Brighina F., Di Stefano V.

Department of Biomedicine, Neuroscience and advanced Diagnostic (BIND), University of Palermo - Palermo - Italy

P41 - LONG-TERM SAFETY, TOLERABILITY, AND EFFICACY OF SUBCUTANEOUS EFGARTIGIMOD PH20 IN PARTICIPANTS WITH GENERALIZED MYASTHENIA GRAVIS: INTERIM RESULTS OF THE ADAPT-SC+ STUDY

Howard Jr J.F.^[1], Antozzi C.^[2], Li Y.^[3], Vu T.^[4], Korobko D.^[5], Steeland S.^[6], Van Hoorick B.^[6], Podhorna J.^[6], Hodari M.^[6], Utsugisawa K.^[7], Saccà F.^[8], Wiendl H.^[9], De Bleecker J.L.^[10], Mantegazza R.^[11]

[1]Department of Neurology, The University of North Carolina - Chapel Hill - United States of America, [2]Fondazione IRCCS Istituto Neurologico "Carlo Besta" - Milan - Italy, [3]Neuromuscular Center, Cleveland Clinic - Cleveland - United States of America, [4]Department of Neurology, University of South Florida Morsani College of Medicine - Tampa - United States of America, [5]State Budgetary Healthcare Institution

of Novosibirsk Region "State Novosibirsk Regional Clinical Hospital" - Novosibirsk - Russian Federation, [6] argenx - Ghent - Belgium, [7]Department of Neurology, Hanamaki General Hospital - Hanamaki - Japan, [8] NRSO Department, Federico II University of Naples - Naples - Italy, [9]Department of Neurology, University of Münster - Münster - Germany, [10]Department of Neurology, Ghent University Hospital - Ghent - Belgium, [11]Department of Neuroimmunology and Neuromuscular Diseases, IRCCS Foundation "Carlo Besta" Neurological Institute - Milan - Italy

P42 - ACHIEVEMENT OF MINIMAL SYMPTOM EXPRESSION IN PARTICIPANTS TREATED WITH EFGARTIGIMOD IN ADAPT+ AND ADAPT-SC+

Hoffman S.^[1], Fionda L.^[2], Muppidi S.^[3], Howard Jr J.F.^[4], Murai H.^[5], Phillips G.^[6], Qi C.^[6], Gelinas D.^[6], Brauer E.^[6], Zhao S.^[6], Bril V.^[7], Vissing J.^[8], Verschuuren J.^[9], Mantegazza R.^[10]

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P43 - ARGX-119 IN PARTICIPANTS WITH DOK7 CONGENITAL MYASTHENIC SYNDROMES: A PHASE 1B STUDY DESIGN

Lochmuller H.^[5], Maggi L.^[1], Burden S.J.^[2], Avendaño J.^[3], Van Bragt T.^[3], Gelinas D.^[3], Dincq S.^[3], Sumbul A.^[3], Vanhuwaert R.^[3], Shilling R.^[3], Palace J.^[4]

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P44 - EFGARTIGIMOD AS A RESCUE THERAPY IN REFRACTORY MYASTHENIC CRISIS

Milella G., Lasorella P., Sozzo M., Scannicchio S., Carlone S., Guglielmi M., Lisi L., Lapenna F.

Department of Translational Biomedicine and Neurosciences (DiBrain), University of Bari Aldo Moro - Bari - Italy

P45 - EFGARTIGIMOD IN PATIENTS WITH GENERALIZED MYASTHENIA GRAVIS UNRESPONSIVE TO OR INTOLERANT OF IVIG

D'Amico E., Campo S., Quartetti U., Portera V., Rini N., Alonge P., Brighina F., Di Stefano V.

Department of Biomedicine, Neuroscience and advanced Diagnostic (BIND), University of Palermo, - Palermo - Italy

P46 - EARLY EFFECT OF INNOVATIVE THERAPIES IN GENERALIZED MYASTHENIA GRAVIS IN THE FIRST WEEK OF TREATMENT

Di Stefano V.^[2], Vanoli F.^[1], Frangiamore R.^[1], Bonanno S.^[1], Antozzi C.^[1], Rodolico C.^[3], Iorio R.^[4], Fionda L.^[5], Massa R.^[6], Sgarzi M.^[7], Maestri M.^[8], Habetswallner F.^[9], Biseco A.^[10], Damato V.^[11], Grandis M.^[12], Saccani E.^[13], Codeluppi L.^[14], Falzone Y.^[15], Liguori R.^[16], Giannoccaro M.P.^[16], Volonghi I.^[17], Alboini P.E.^[18], Tonin P.^[19], Cheli M.^[19], Rigamonti A.^[20], Vinciguerra C.^[21], Rini N.^[2], Giacopuzzi Grigoli E.^[1], D'Alvano G.^[22], Brighina F.^[2], Maggi L.^[1]

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Cuore, Roma - Roma - Italy, [5]Dipartimento di Neurologia, Ospedale Sant'Andrea Roma - Roma - Italy, [6] Dipartimento Neurologia Policlinico Tor Vergata di Roma - Roma - Italy, [7]Dipartimento di Neurologia, Ospedale Papa Giovanni XXIII di Bergamo - Bergamo - Italy, [8]Azienda Universitaria Ospedaliera di Pisa - Pisa - Italy, [9]A.O. Antonio Cardarelli di Napoli - Napoli - Italy, [10]A.O.U Università della Campania Luigi Vanvitelli - Napoli - Italy, [11]A.O.U. Careggi di Firenze - Firenze - Italy, [12]Policlinico San. Martino di Genova - Genova - Italy, [13]A.O.U. di Parma - Parma - Italy, [14]UO Neurologia, AUSL IRCCS di Reggio Emilia - Reggio Emilia - Italy, [15]UO Neurologia, Ospedale San Raffaele di Milano - Milano - Italy, [16]IRCCS Istituto delle Scienze Neurologiche di Bologna, Bologna - Bologna - Italy, [17]Dipartimento di Continuità di Cura e Fragilità SC Neurologia ASST Spedali Civili di Brescia - Brescia - Italy, [18]UOC Neurologia Fondazione IRCCS Casa Sollievo della Sofferenza San Giovanni Rotondo (FG) - San Giovanni Rotondo - Italy, [19]Neurologia B, Azienda Ospedaliera Universitaria Integrata Verona - Verona - Italy, [20]UOC Neurologia, ASST Lecco - Lecco - Italy, [21]Ospedale Universitario di Salerno - Salerno - Italy, [22]Department of Advanced Medical and Surgical Sciences-University of Campania Luigi Vanvitelli, Italy - Napoli - Italy

P47 - ECULIZUMAB AS TREATMENT IN REFRACTORY IMPENDING AND MYASTHENIC CRISIS: A CASE SERIES

Ricciardi D., Erra C., Matrone F., Fasolino A., Tuccillo F., Habetswallner F.

AORN Cardarelli - UOC Clinical Neurophysiology - Napoli - Italy

P48 - EFGARTIGIMOD IN MYASTHENIA GRAVIS: A REAL LIFE SINGLE-CENTER EXPERIENCE

Erra C.^[1], Fasolino A.^[1], Signori A.^[2], Ricciardi D.^[1], Francesco T.^[1], Federica M.^[1], Francesco H.^[1]

[1]AORN A. Cardarelli - Napoli - Italy, [2]University of Genoa, Department of Health Sciences, Section of Biostatistics - Genoa - Italy

P49 - PROMINENT AND FAST RESPONSE TO ECULIZUMAB IN MYASTHENIC CRISIS: THE POTENTIAL AS RESCUE THERAPY IN REFRACTORY MYASTHENIA GRAVIS

Messina C.^[1], Basile L.^[2], Crescimanno G.^[1], Battaglia S.^[2], Scichilone N.^[2], Brighina F.^[1], Di Stefano V.^[1]

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P50 - MALIGNANCY RISK AND AZATHIOPRINE IN MYASTHENIA GRAVIS: A RETROSPECTIVE COHORT STUDY FROM A SINGLE CENTER

Drago S.F.A., Pugliese A., Biasini F., Barbaccia A., Luppino F., Musumeci O., Rodolico C.

Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy - Messina - Italy

P51 - HIGH DOSE INTRAVENOUS METHYLPREDNISOLONE THERAPY FOR OCULAR MYASTHENIA GRAVIS. A FEASIBILITY AND SAFETY STUDY.

Cortese F.^[1], Fionda L.^[2], Antonini G.^[3], Morino S.^[2], Rossini E.^[3], Brienza M.^[1], De Giglio L.^[1], Altavista M. C.^[4], Pennisi E. M.^[1]

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P52 - COMPLEMENT-INHIBITORS FOR MYASTHENIC CRISIS: A PROMISING AND FAST-ACTING THERAPY

Vera V., Rossini E., Morino S., Garibaldi M., Leonardi L., Tufano L., Lauletta A., Forcina F., Antonini G., Fionda L.

Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome - Rome - Italy

P53 - COMPLEMENT INHIBITOR THERAPY IN THYMOMA-ASSOCIATED MYASTHENIA GRAVIS: A REAL-WORLD EXPERIENCE

Marini S.^[2], Erra C.^[1], Fionda L.^[3], Falso S.^[2], Rossini E.^[3], Habetzwallner F.^[2], Meacci E.^[4], Marini M.^[2], Habetzwallner F.^[1], Iorio R.^[5]

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P54 - COMPLEMENT INHIBITION IN MYASTHENIA GRAVIS: A REAL WORLD EXPERIENCE WITH RAVULIZUMAB.

Frangiamore R.^[1], Vanoli F.^[1], Bonanno S.^[1], Maggi L.^[1], Eleonora G.G.^[1], Cheli M.^[1], Canella M.^[2], Cavalcante P.^[1], Iacomino N.^[1], Rinaldi E.^[1], Tarasco M.C.^[1], Berni A.^[1], Mantegazza R.E.^[3], Antozzi C.G.^[1]

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Myasthenia Gravis II P55 - P73



Friday, May 30, 2025 15:45 - 17:30

Chairpersons: Antonio Petrucci (Roma), Dario Ricciardi (Napoli)

P55 - ITALIAN SINGLE-BREATH COUNT TEST: RETROSPECTIVE ANALYSIS IN A SINGLE-CENTRE COHORT OF MYASTHENIA GRAVIS PATIENTS

Nuredini A.^[1], Libelli G.^[1], D'Orsi A.^[1], Chierici E.^[2], Allegri I.^[2], Saccani E.^[2]

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P56 - ANTI-FCRN TREATMENT WITH ROZANOLIXIZUMAB IN GENERALIZED MYASTHENIA GRAVIS: A REAL WORD EXPERIENCE

Frangiamore R.^[1], Canella M.^[2], Vanoli F.^[1], Bonanno S.^[1], Maggi L.^[1], Cheli M.^[1], Giacomuzzi Grigoli E.^[1], Mantegazza R.E.^[3], Antozzi C.G.^[1]

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P57 - EXPLORING THE ROLE OF ENDOPLASMIC RETICULUM STRESS RESPONSE IN NEUROMUSCULAR JUNCTION IMPAIRMENT IN MYASTHENIA GRAVIS: A POSSIBLE NOVEL CONTRIBUTOR TO AUTOANTIBODY-INDUCED ANTIGENIC MODULATION

Iacomino N., Giagnorio E., Tarasco M.C., Berni A., Malacarne C., Frangiamore R., Marcuzzo S., Antozzi C., Mantegazza R., Cavalcante P.

Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta - Milano - Italy

P58 - A RETROSPECTIVE COHORT MULTICENTER STUDY ON SAFETY AND EFFECTIVENESS OF MYCOPHENOLATE MOFETIL IN GENERALIZED MYASTHENIA GRAVIS

Bevilacqua L.^[1], Di Stefano V.^[2], D'Amico A.^[2], Rini N.^[2], D'Apolito M.^[3], Liberatoscioli E.^[3], Monastero R.^[2], Brighina F.^[2], Barone P.^[4], Vinciguerra C.^[1]

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P59 - MYASTHENIA GRAVIS AND MYELOID NEOPLASMS: A RECURRENT ASSOCIATION WITH A POSSIBLE SHARED THERAPY

Habetzwallner F.^{*[1]}, Falso S.^[1], Marini S.^[1], Marini M.^[1], Torchia E.^[1], Monforte M.^[2], Ravera B.^[1], Bortolani S.^[2], Ricci E.^[2], Iorio R.^[2]

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P60 - THE IMPORTANCE OF NEUROPHYSIOLOGY TESTING IN SERONEGATIVE MYASTHENIA GRAVIS: A RETROSPECTIVE STUDY IN TWO TERTIARY CENTERS IN SOUTHERN ITALY

Rini N.^[1], Maccora S.^[1], Vinciguerra C.^[2], Messina C.^[1], Alonge P.^[1], Brighina F.^[1], Di Stefano V.^[1]

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P61 - EFFICACY OF RAVULIZUMAB IN A PATIENT WITH MYASTHENIA GRAVIS AND MYH7-RELATED MYOPATHY

Urbano G.^[1], Gadaleta G.^[1], Pidello S.^[2], Deaglio S.^[5], Tucci R.^[3], Morra M.^[3], Vigliani M.C.^[4], Mongini T.E.^[1], Vercelli L.^[1]

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P62 - RISK OF FALSE ACETYLCHOLINE RECEPTOR AUTOANTIBODY POSITIVITY TESTING BY RADIOIMMUNOPRECIPITATION ASSAY

Zara P.^[1], Chessa P.^[1], Deiana G.A.^[1], Morette A.^[1], Puci M.^[1], Sotgiu G.^[1], Solla P.^[1], Damato V.^[2], Sechi E.^[1]

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P63 - A POTENTIAL SYNERGISTIC ROLE OF AIRE AND HLA IN MYASTHENIA GRAVIS PATHOGENESIS

Marini M., Falso S., Marini S., Morroni J., Valenzano G., Morcone V., Habetswallner F., Iorio R.
Fondazione Agostino Gemelli - Roma - Italy

P64 - EXPLORING THE ASSOCIATION BETWEEN HEALTH LITERACY AND DAILY FUNCTIONING IN PATIENTS AFFECTED BY MYASTHENIA GRAVIS: A CROSS-SECTIONAL STUDY

Pastore F.^[1], Forastefano B.^[2], Milella G.^[3], Minoia F.^[2], Domenicone E.^[4], Lapenna F.^[3], Simonetti V.^[5], Cicolini G.^[5], Comparcini D.^[6]

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P65 - THE ROLE OF IMMUNOADSORPTION IN TREATING MYASTHENIA GRAVIS EXACERBATIONS IN THE ERA OF MOLECULAR TARGET THERAPIES: A CASE DESCRIPTION.

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P66 - ROZANOLIXIZUMAB IN TREATMENT OF GENERALIZED MYASTHENIA GRAVIS WITH ACHR-AB: A SINGLE CENTER CASE SERIES

Cepele A.^[1], Guida M.^[1], Iovino V.^[1], Latini E.^[1], Sozzo M.^[2], Siciliano G.^[1], Maestri Tassoni M.^[1]

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P67 - MYASTHENIA GRAVIS, BEYOND THE NEUROMUSCULAR JUNCTION: AN EEG-BASED FUNCTIONAL CONNECTIVITY PILOT STUDY

Greco G.^[2], Conti M.^[1], Frezza E.^[2], Goglia M.^[2], Nardino G.^[2], Petitta I.^[2], Gruosso F.^[2], Vietri G.^[2], Boffa L.^[2], Stefani A.^[1], Massa R.^[2]

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P68 - EXPLORING THE CORRELATION BETWEEN FATIGUE, DEPRESSION, SLEEP DISORDERS AND CLINICAL SEVERITY IN MG PATIENTS

Greco G., Frezza E., Goglia M., Nardino G., Petitta I., Gruosso F., Vietri G., Boffa L., Massa R.
Neuromuscular Diseases Unit, Policlinico e Università degli Studi di Roma Tor Vergata - Rome - Italy

P69 - NEW THERAPEUTIC OPPORTUNITIES IN MYASTHENIA GRAVIS

Masuzzo F., Maglione A., Torrieri M.C., Virgilio E., Ferri R., Clerico M.
SSD Patologie Neurologiche Specialistiche - Orbassano (TO) - Italy

P70 - ARE PEDIATRIC MYASTHENIAS TRULY LESS COMPLEX THAN ADULT MYASTHENIAS? INSIGHTS FROM 15 YEARS OF EXPERIENCE IN A NEUROMUSCULAR TERTIARY CARE CENTER

Gallo G.^[1], D'alessandro R.^[1], Cavallina I.^[1], Rolle E.^[1], Buscaglia A.^[1], Montin D.^[2], Mongini T.^[3], Ricci F.^[1]

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P71 - THYMOMA-ASSOCIATED ANTI-MUSK MYASTHENIA GRAVIS AND ANTI-CASPR2 PARANEOPLASTIC ENCEPHALITIS: A CASE REPORT OF A VERY RARE ASSOCIATION

Iovino V.^[1], Maestri Tassoni M.^[1], Cepele A.^[1], Latini E.^[1], Sozzo M.^[2], Siciliano G.^[1], Guida M.^[1]

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P72 - MAGNETIC RESONANCE IMAGING REVEALING BULBAR MUSCLE INVOLVEMENT IN ACHR-POSITIVE MYASTHENIA GRAVIS: A CASE REPORT

Marando D.^[1], Leonardi L.^[2], Garibaldi M.^[1], Morino S.^[2], Rossini E.^[1], Lauletta A.^[1], Antonini G.^[1], Fionda L.^[2]

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P73 - IDENTIFICATION OF CLINICAL VARIABLES ASSOCIATED WITH TREATMENT RESPONSE TO EFGARTIGIMOD IN GENERALIZED MG

Vanoli F., Frangiamore R., Bonanno S., Giacomuzzi Grigoli E., Cheli M., Maggi L., Antozzi C.

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Duchenne Muscular Dystrophy P74- P99

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: Luisa Politano (Napoli), Michela Catteruccia (Roma)

P74 - FAT EMBOLISM AFTER FRACTURES IN DUCHENNE MUSCULAR DYSTROPHY: AN UNDERDIAGNOSED COMPLICATION

Tosi M.^[1], Catteruccia M.^[1], Diodato D.^[1], Adorisio R.^[2], Perrotta D.^[3], D'Amico A.^[1]

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P75 - LONG-TERM EFFICACY, SAFETY, AND MICRO-DYSTROPHIN EXPRESSION AFTER DELANDISTROGENE MOXEPARVOVEC TREATMENT IN DUCHENNE: 2-YEAR FOLLOW-UP OF EMBARK STUDY (NCT05096221)

Mercuri E.M.^[1], Mendell J.R.^[11], Muntoni F.^[2], Mcdonald C.M.^[3], Ciafaloni E.^[1], Komaki H.^[4], Leon-Astudillo C.^[5], Nascimento A.^[6], Proud C.^[7], Schara--Schmidt U.^[8], Veerapandian A.^[9], Zaidman C.M.^[10], Furgerson M.^[11], Ding K.^[11], Singh P.^[11], Potter R.^[11], Asher D.R.^[11], Murphy A.P.^[12], Reid C.^[12], Hooper G.^[12], Torre C.O.^[12], Manfrini M.^[13], Elkins J.S.^[11], Rodino-Klapac L.R.^[11]

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P76 - MODELING DISEASE PROGRESSION IN DUCHENNE MUSCULAR DYSTROPHY: REDUCED DECLINE IN FORCED VITAL CAPACITY WITH GIVINOSTAT COMPARED WITH STANDARD OF CARE

Finanger E.L.^[3], Lavery C.G.^[1], Alessi F.^[2], Cazzaniga S.^[2], Bettica P.^[2], Largajolli A.^[4], Bellanti F.^[4], Péréon Y.^[5], Mcdonald C.M.^[6], Mercuri E.M.^[7]

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P77 - 3-YEAR FUNCTIONAL OUTCOMES OF PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY: POOLED DELANDISTROGENE MOXEPARVOVEC CLINICAL TRIAL DATA VS EXTERNAL CONTROLS

Bruno C.^[1], Connolly A.M.^[2], Day J.^[3], Mcdonald C.^[4], Proud C.^[5], Shieh P.^[6], Zaidman C.^[7], Ferguson M.^[8], Ding K.^[8], Reid C.^[9], Murphy A.P.^[9], Elkins J.S.^[8], Rodino-Klapac L.^[8], Mendell J.^[8]

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P78 - PERSONALIZED AEROBIC TRAINING IN DUCHENNE MUSCULAR DYSTROPHY (DMD): AN EXPLORATORY STUDY TO EVALUATE SAFETY, FEASIBILITY, AND IMPACT ON FATIGUE AND QUALITY OF LIFE.

Branca A.^[1], Mangone S.^[1], Briganti M.^{[1][3]}, Onofri M.^[1], Bruschini M.^[2], Sancricca C.^{[1][3]}, Frongia A.L.^{[1][4]}

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P79 - DISEASE PROGRESSION MODELING IN DUCHENNE MUSCULAR DYSTROPHY: DELAYED DECAY IN 4-STAIR CLIMB WITH GIVINOSTAT COMPARED WITH STANDARD OF CARE

Lavery C.G.^[1], Mercuri E.M.^[2], Alessi F.^[3], Cazzaniga S.^[3], Bettica P.^[3], Largajolli A.^[4], Bellanti F.^[4], Péron Y.^[5], Mcdonald C.M.^[6], Finanger E.L.^[7]

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P80 - GIVINOSTAT EFFECT ON RESPIRATORY FUNCTION IN DUCHENNE MUSCULAR DYSTROPHY BEFORE AND AFTER AMBULATION LOSS: RESULTS FROM EPIDYS, LTSE, AND PRO-DMD-01 STUDIES

Mcdonald C.M.^[1], Lavery C.G.^[2], Finanger E.L.^[3], Signorovitch J.E.^[5], Parodi A.^[4], Alessi F.^[4], Cazzaniga S.^[4], Bettica P.^[4], Péron Y.^[6], Mercuri E.M.^[2]

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P81 - ALTERNATIVE TRANSCRIPT ISOFORMS IN PREDICTING DMD-RELATED PHENOTYPE: A PRELIMINARY STUDY

Risi B.^[1], Giacometti L.^[2], Corsini M.^[3], Romani C.^[4], Piloni C.^[5], Bugatti M.^[5], Caria F.^[6], Damioli S.^[6], Poli L.^[8], Baronchelli C.^[7], Padovani A.^[9], Mitola S.^[3], Filosto M.^[10], Lanzi G.^[11]

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P82 - EFFICACY OF GIVINOSTAT IN THE OFF-TARGET POPULATION OF EPIDYDS: A SUBGROUP ANALYSIS

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P83 - JMW2894, A GROWTH HORMONE SECRETAGOGUES (GHS): EFFECTS ON AN IN VITRO MODEL OF DUCHENNE MUSCULAR DYSTROPHY

Bresciani E.^[1], Meanti R.^[1], Rizzi L.^[1], Sartori E.^[1], Bassani G.^[1], Liantonio A.^[2], Cappellari O.^[2], De Luca A.^[2], Fehrentz J.^[3], Denoyelle S.^[3], Torsello A.^[1]

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P84 - EVALUATION OF BEHAVIOURAL PROBLEMS USING PARS III IN THE VISION-DMD STUDY OF VAMOROLONE VS PREDNISONE IN DUCHENNE MUSCULAR DYSTROPHY (DMD)

Henricson E.^[2], De Vera A.^[3], Nip K.^[3], Clemens P.R.^[7], Guglieri M.^[4], Truba N.^[5], D'Angelo M.G.^[1], Hoffman E.P.^[6]

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P85 - COMPARISON BETWEEN D-LOOP METHYLATION LEVEL AND MTDNA COPY NUMBER IN DUCHENNE AND BECKER MUSCULAR DYSTROPHIES PATIENTS

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P86 - BONE HEALTH AND RISK OF VERTEBRAL FRACTURE IN DMD:

WHICH ROLE THE FUNCTIONAL STATUS PLAYS?

Arpaia C.^[1], Capasso A.^[1], Villa M.^[1], Panicucci C.^[2], Repetto A.^[2], Coratti G.^[1], Gulli C.^[3], Marzoli A.^[4], Lala M.R.^[1], Pane M.^[1], Di Iorgi N.^[5], Bruno C.^[2], Mercuri E.^[1]

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P87 - LONGITUDINAL ANALYSIS OF MOTOR FUNCTION DECLINE IN DUCHENNE MUSCULAR DYSTROPHY: A COHORT STUDY.

Gardani A.^[1], Iosca A.^[2], Dainesi M.I.^[2], Khalil S.^[2], Carraro L.^[2], Berardinelli A.L.^[1]

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P88 - ROLLING TIME IN DMD: DATA FROM A LARGER COHORT CONFIRM OUR PREVIOUS ANALYSIS

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P89 - OPTICAL GENOME MAPPING: VALUABLE DIAGNOSTIC TOOL FOR DMD FEMALE CARRIERS

Alesi V.^[4], Fattori F.^[4], Bosco L.^[4], Mietto M.^[4], Catino G.^[1], Ferlini A.^[2], Bertini E.^[3], Novelli A.^[4], D'Amico A.^[3]

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P90 - THE PNRR NANOTHER PROJECT: TESTING NOVEL NANODELIVERY OF RNA MOLECULES AND SYNTHETIC GENES AS THERAPEUTIC APPROACHES FOR DUCHENNE MUSCULAR DYSTROPHY

Montanari S.^[1], Mietto M.^[1], Falzarano M.S.^[2], Pinton P.^[2], Giorgi C.^[2], Cavalli R.^[3], Argenziano M.^[3], Tedesco F.S.^[4], Ferlini A.^[1]

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P91- PRE-CLINICAL ASSESSMENT OF THE ROLE OF THE COMPLEMENT SYSTEM IN TRIGGERING FIBROSIS IN DUCHENNE MUSCULAR DYSTROPHY: TOWARDS NOVEL THERAPEUTIC TARGETS

Tulimiero L., Mantuano P., Lenti R., Quarta R., Cristiano E., Marinelli M., Boccanegra B., Cappellari O., De Luca A.

Section of Pharmacology, Department of Pharmacy - Drug Sciences, University of Bari "Aldo Moro", Bari, Italy - Bari - Italy

P92 - IDENTIFICATION AND TREATMENT OF NEURODEVELOPMENTAL AND MENTAL DISORDERS IN BOYS AND ADULTS WITH DUCHENNE MUSCULAR DYSTROPHY: A COHORT STUDY

Malaspina M., Brogna C., Moriconi F., Capasso A., Arpaia C., Cicala G., Ricci M., Villa M., Pellizzari M., De Gioia A., Falco A., Turano M., Coratti G., Janiri D., Sani G., Chieffo D.P.R., Pane M., Mercuri E.M.

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P93 - DIAGNOSIS OF DUCHENNE MUSCULAR DYSTROPHY IN ITALY OVER THE LAST DECADE: HAVE WE MADE PROGRESS?

Tosi M.^[1], Panicucci C.^[2], Paolucci S.^[3], Magri F.^[4], Giannotta M.^[5], Masson R.^[6], Albamonte E.^[7], Bruno I.^[8], Filosto M.^[9], Zambon A.^[10], Picillo E.^[11], Politano L.^[12], Berardinelli A.^[13], Pera M.C.^[14], Sframeli M.^[15], Ricci F.^[16], Bello L.^[17], Bruno C.^[2], Catteruccia M.^[1], Mercuri E.^[3], Pini A.^[5], Sansone V.^[7], Previtali S.^[10], Nigro V.^[11], Messina S.^[15], Pegoraro E.^[17], Bertini E.^[1], D'Amico A.^[1]

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P94 - STORE-OPERATED CALCIUM ENTRY: A NOVEL PHARMACOLOGICAL TARGET FOR DUCHENNE MUSCULAR DYSTROPHY

Muschiatiello C.^[1], Azzarone L.^[1], Pessolano E.^[2], Gallo E.^[2], Reano S.^[3], Filigheddu N.^[3], Piralì T.^[2], Panicucci C.^[4], Bruno C.^[4], Riva B.^[1]

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P95 - CARDIAC MANAGEMENT IN DUCHENNE MUSCULAR DYSTROPHY: ASSESSING THE IMPACT OF EARLY PROPHYLAXIS. A RETROSPECTIVE SINGLE-CENTER STUDY

Giannotta M.^[1], Di Tella B.^[2], Nardoza M.^[3], Trentin F.^[2], Scarpini G.^[1], Cordelli D.M.^[2], Pini A.^[1]

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P96 - INERTIAL TECHNOLOGY FOR THE MOTOR FUNCTION ASSESSMENT IN NEUROMUSCULAR DISORDERS (NMDS): PROTOCOL DESIGN AND PRELIMINARY RESULTS IN PEDIATRIC PATIENTS.

D'alessandro R.^[1], Elisa P.^[2], Cavallina I.^[1], Sertori F.^[1], Rolle E.^[1], Rossi F.^[1], Gastaldi L.^[2], Mongini T.E.^[3], Ricci F.^[1]

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P97 - PERIPHERAL IMMUNOLOGICAL SIGNATURE OF PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY

Baratto S.^[1], Panicucci C.^[1], Pintus S.^[1], Ferrara I.^[1], Mela F.^[1], Principi E.^[1], Antonini F.^[2], Del Zotto G.^[2], Raffaghello L.^[1], Bruno C.^[1]

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P98 - LONGITUDINAL MOTOR FUNCTIONAL AND MUSCLE MAGNETIC RESONANCE FINDINGS IN A COHORT OF DMD PATIENTS WITH SMALL MUTATIONS

Brogna C.^[1], Cristiano L.^[1], Verdolotti T.^[1], Varcasia G.^[1], Gaudino S.^[1], Mercuri E.^[1], Tasca G.^[2]

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P99 - HOW MANY PATIENTS ARE STILL INVISIBLE?": REAL-WORLD EPIDEMIOLOGICAL STUDY OF DUCHENNE MUSCULAR DYSTROPHY AND SPINAL MUSCULAR ATROPHY IN TUSCANY

Ricci G.^[1], Rende M.^[1], Loprieno S.^[1], Torri F.^[1], Sacchini M.^[2], Ticci C.^[2], Procopio E.^[2], Astrea G.^[3], Battini R.^[3], Santoro M.^[4], Coi A.^[4], Pieroni F.^[4], Francesconi P.^[5], Siciliano G.^[1]

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Myotonic Dystrophies - Muscular Dystrophies

P100 - P121

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: Cristina Sancricca (Roma), Grazia Crescimanno (Palermo)

P100 - NEW MODELS FOR Δ -SARCOGLYCANOPATHY: A COMBINED ZEBRAFISH AND CELL-BASED APPROACH

Dalla Barba F.^[1], Scano M.^[1], Benetollo A.^[1], Akyurek E.E.^[2], Carotti M.^[1], Dorianna S.^[1], Argenton F.^[3]

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P101- PATIENT-REPORTED OUTCOME MEASURES IN NUCLEAR ENVELOPE MYOPATHIES IN A LARGE COHORT OF PATIENTS

Cheli M.^[1], Cugudda E.^[2], Kristjánsdóttir S.M.^[3], Maggi L.^[1]

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P102 - HOMOZYGOUS P.(ILE309SER) VARIANT IN THE SNUPTIN 1 GENE IN A NOVEL CASE OF LGMDR29.

Magri F.^[1], Rimoldi M.^[2], Velardo D.^[1], Gagliardi D.^[3], Arima G.M.^[4], Zanotti S.^[1], Napoli L.^[1], Ripolone M.^[1], Ciscato P.^[1], Bertolasi L.^[1], Moliterno N.^[4], Sciacco M.^[1], Comi G.P.^[4], Corti S.^[4], Ronchi D.^[4]

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P103 - THE P.ASP753ASN VARIANT IN CALPAIN-3: STRUCTURAL AND FUNCTIONAL IMPLICATIONS IN AUTOSOMAL DOMINANT LIMB-GIRDLE MUSCULAR DYSTROPHY

D'Este G.^[1], Cassandrini D.^[2], Magri F.^[3], Ronchi D.^[5], Rubegni A.^[4], Vattemi G.^[6], Tonin P.^[6], Lopercolo D.^[7], Malandrini A.^[7], Merlini L.^[8], Barresi R.^[1]

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P104 - COGNITIVE AND BEHAVIORAL PROFILE IN PEDIATRIC MYOTONIC DYSTROPHY: LITERATURE OVERVIEW AND REAL WORLD EXPERIENCE

Pozzi S.^[1], Albamonte E.^[1], Casiraghi J.^[1], Gaudet I.^[2], Angeard N.^[3], Nikolenko N.^[4], Ekstrom A.^[5], Braakman H.^[6], Hendriksen J.G.M.^[7], Bassez G.^[8], Gourdon G.^[9], Pereira M.G.^[9], Wang E.^[10], Trucco F.^[11], Kornblum C.^[12], Sweere D.^[13], Eisengart J.^[14], Berggren K.^[15], Johnson N.^[15], Sansone V.A.^[1]

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P105 - BEYOND CARDIOMYOPATHY: A CASE OF CORONARY ARTERY DISEASE IN BECKER MUSCULAR DYSTROPHY

Palermo C.^[1], Leone D.^[1], Berti B.^[1], Lamendola P.^[2], Amore L.^[2], Lombardo A.^[2], Lanza G.A.^[3], Mercuri E.^[4], Trani C.^[3], Burzotta F.^[3], Pane M.^[4]

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P106 - A LONG WAY DOWN: RETROSPECTIVE REVIEW OF TRACHEOTOMIES IN A POPULATION OF DM1 PATIENTS FOLLOWED AT THE NEMO CLINICAL CENTER IN MILAN.

Ferrari Aggradi C.R.^{[1][2]}, Gotti E.^[2], Zanolini A.^[2], Colacicco G.^[2], Roma E.^[2], Casiraghi J.^[2], Sansone V.A.^[2]

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P107 - IDENTIFICATION, CHARACTERIZATION AND SEGREGATION ANALYSIS OF DM1 ALLELES CONTAINING NON-CTG VARIANT REPEATS (VRS) IN A TWO-GENERATION ITALIAN FAMILY

Visconti V.V.^[1], Centofanti F.^[1], D'Apice M.R.^[2], Ciabini D.^[1], Carlomagno M.^[3], Maestri S.^[3], Esposito A.^[3], Rossato M.^[3], Delledonne M.^[3], Goglia M.^[4], Greco G.^[4], Frezza E.^[4], Massa R.^[4], Botta A.^[1], Novelli G.^[1]

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P108 - NEW METHOD TO IDENTIFY MYOTONIC DYSTROPHY TYPE 2 PATIENTS CARRYING VARIANT (TCTG)N REPEATS AT THE 3' ENDS OF THE CNBP EXPANDED ALLELES.

Centofanti F.^[1], Visconti V.V.^[1], D'Apice M.R.^[2], Turchetti E.^[1], Ciabini D.^[1], Bagni I.^[2], Rossato M.^[3], Carlomagno M.^[3], Maestri S.^[3], Delledonne M.^[3], Goglia M.^[4], Greco G.^[4], Frezza E.^[4], Massa R.^[4], Botta A.^[1], Novelli G.^[1]

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P109 - CELL LINES OF DUCHENNE MUSCULAR DYSTROPHY PATIENTS SHOWS MUTATION-DEPENDENT DELAY IN MYOGENIC PROGRAM

Marinelli M.^[2], Cristiano E.^[2], Quarta R.^[2], Boccanegra B.^[2], Cerchiara A.G.^[2], Barile S.^[3], Mouly V.^[1], Lasorsa F.M.^[3], Imbriani P.^[2], Cappellari O.^[2], De Luca A.^[2]

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P110 - GENETIC AND CLINICAL SPECTRUM OF LIMB-GIRDLE MUSCULAR DYSTROPHIES IN WESTERN SICILY: A SINGLE-CENTER COHORT STUDY

Rini N.^[1], Alonge P.^[1], Lupica A.^[1], Borgione E.^[2], Scuderi C.^[2], Santapaola S.^[2], Lo Giudice M.^[2], Brighina F.^[1], Di Stefano V.^[1]

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P111 - NATURAL HISTORY OF HAND MUSCLE WEAKNESS PROGRESSION IN MYOTONIC DYSTROPHY TYPE 1 (DM1)

Goglia M., Frezza E., Greco G., Petitta I., Gruosso F., Vietri G., Nardino G., Boffa L., Massa R.

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P112 - PREVALENCE OF MENINGIOMA IN MYOTONIC DYSTROPHY TYPE 1

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P113 - LIMB-GIRDLE MUSCULAR DYSTROPHIES: CLINICAL CHARACTERISTICS OF LGMD2A/R1 AND LGMD2B/R2 PATIENTS FROM A SINGLE ITALIAN CENTRE

Luppino F., Musumeci O., Pugliese A., Materia R., La Rosa E., Lauro R., Said I., Allegra C., Toscano A., Rodolico C., Messina S., Sframeli M.

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P114 - A SEVERE FORM OF ENDOMETRIOSIS IN A YOUNG WOMAN WITH STEINERT DISEASE

Loprieno S.^[1], Vadi G.^[1], Iaquina G.^[1], Torri F.^[2], Marciano E.^[3], Santoro A.^[4], Carpenè N.^[5], Ricci G.^[1], Siciliano G.^[1]

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P115 - PEDIATRIC MUSCULAR DYSTROPHIES: EXPLORING THE CORRELATION BETWEEN MOTOR FUNCTION AND MUSCLE IMAGING IN AMBULANT PATIENTS. PRELIMINARY DATA FROM A SINGLE-CENTER RETROSPECTIVE STUDY

Trentin F.^[1], Lambertini E.^[2], Maffei M.^[3], Giannotta M.^[2], Scarpini G.^[2], Briganti E.^[1], Not R.^[4], Lodi R.^[4], Cordelli D.M.^[1], Pini A.^[2]

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P116 - A CASE SERIES OF LGMD R1 CALPAIN 3-RELATED PATIENTS PRESENTING THE HYPOMORPHIC VARIANT C.1746-20C>G

Magri F.^[1], Berardinelli A.^[2], Capece G.^[3], Gagliardi D.^[4], Villella C.^[5], Velardo D.^[1], Zanotti S.^[1], Ciscato P.^[1], Molitierno N.^[4], Fortunato F.^[5], Sciaccio M.^[1], Ricci G.^[6], Siciliano G.^[6], Bello L.^[3], Pegoraro E.^[3], Giacomo C.^[5], Corti S.^[5], Ronchi D.^[5]

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P117 - DILATED CARDIOMYOPATHY ONSET AND PROGRESSION IN THE BECKER MUSCULAR DYSTROPHY COHORT FOLLOWED AT THE UNIVERSITY OF PADOVA

Petrosino A.^[1], Riguzzi P.^[1], Capece G.^[1], Sogus E.^[1], Gorgoglione D.^[1], Sabbatini D.^[2], Vianello S.^[1], Calore C.^[2], Bauce B.^[2], Pegoraro E.^[1], Bello L.^[1]

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P118 - A FAMILY WITH DISTAL MYOPATHY CARRYING HETEROZYGOUS MUTATIONS IN THE DES AND TK2 GENES WITHOUT A CLEAR GENOTYPE-PHENOTYPE CORRELATION

Vadi G.^[1], Lopriore P.^[1], Lopriore S.^[1], Torri F.^[1], Rende M.^[2], Ciurli B.^[2], Mancuso M.^[1], Siciliano G.^[1], Ricci G.^[1]

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P119 - SENSORY AXONAL NEUROPATHY IN A PATIENT WITH ANOCTAMIN-5 (ANOS)-RELATED LIMB-GIRDLE MUSCULAR DYSTROPHY R12

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P120 - EDINBURGH COGNITIVE AND BEHAVIOURAL ALS SCREEN (ECAS) IN MYOTONIC DYSTROPHIES: A VALUABLE SCREENING TOOL FOR COGNITIVE IMPAIRMENT.

Petitta I.^[1], Frezza E.^[1], Goglia M.^[1], Carmela S.^[2], Greco G.^[1], Gruosso F.^[1], Vietri G.^[1], Boffa L.^[3], Nuccetelli M.^[2], Bernardini S.^[2], Massa R.^[1]

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P121 - BRIDGING GENETIC UNCERTAINTY WITH MUSCLE BIOPSY: A CASE REPORT OF TWO SIBLINGS WITH CALPAINOPATHY AND TURNER SYNDROME

Bruno G.^[1], Ruggiero L.^[2], Rubino A.^[1], Nigro V.^[3], Varone A.^[1]

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Basic Myology - Genetics - Congenital Myopathies

P122- P138

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: *Alessandra Ferlini (Ferrara), Corrado Angelini (Padova)*

P122 - A NOVEL DEEP INTRONIC MUTATION EXPANDS THE GENOTYPE SPECTRUM OF MYH7-RELATED MYOPATHIES

Baro A.^[1], Vivaldi I.^[2], Fortunato F.^[2], Neri M.^[2], Neri L.^[3], Maggi L.^[4], Zuccarino R.^[1], Cenacchi G.^[5], De Iorio M.^[6], Iascone M.^[7], Gualandi F.^[2]

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P123 - EXERCISE INDUCED RHABDOMYOLYSIS IN RRM2B RELATED MYOPATHY

Falcone G.M.I.^[1], Arena I.G.^[1], Drago S.F.A.^[1], Melilli C.^[1], Santorelli F.M.^[2], Musumeci O.^[3]

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P124 - GENETIC NEWBORN SCREENING BY TREAT PANEL TARGETED SEQUENCING IN A LARGE EUROPEAN COHORT: PIPELINE DESIGN AND FIRST RESULTS FROM THE SCREEN4CARE EU PROJECT

Agolini E.^[1], Leslie M.^[2], Einhorn M.^[3], Fortunato F.^[4], Macari G.^[5], Caforio L.^[5], Zuccolo M.^[6], Martinez--Fresno M.^[7], Kirschner J.^[8], Freyler K.^[8], Davoine E.^[9], Faivre L.^[9], Tran--Mau--Them F.^[9], Selvatichi R.^[10], Tonda R.^[2], Morgan C.^[2], Gut I.^[2], Margutti A.^[4], Nagliati V.^[4], Mietto M.^[4], Montanari S.^[4], Sansen S.^[11], Bertini E.^[1], Novelli A.^[1], Beltran S.^[2], Ferlini A.^[4]

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P125 - AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY: A GENETIC SCREENING IN SICILIAN PATIENTS WITH NEURODEVELOPMENTAL AND NEUROMUSCULAR DISORDERS

Scuderi C.^[1], Borgione E.^[1], Lo Giudice M.^[1], Giuliano M.^[1], Pettinato R.^[1], Di Stefano V.^[2], Di Blasi F.D.^[1], Santa Paola S.^[1]

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P126 - TWO NOVEL CASES OF VWA1 NEUROMYOPATHY: CLINICAL AND INSTRUMENTAL DESCRIPTION

Diodato D.^[1], Catteruccia M.^[1], Fattori F.^[2], Bosco L.^[2], Santorelli J.^[1], Tosi M.^[1], D'Amico A.^[1]

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P127 - A NEW FORM OF DISTAL SPINAL MUSCLE ATROPHY RELATED TO NOVEL LOSS-OF-FUNCTION VARIANTS IN ADCY6

Bosco L.^[1], Fattori F.^[1], D'Amico A.^[2], Pro S.^[3], Colona V.L.^[4], Vasco G.^[4], Nicita F.^[2], Novelli A.^[1], Bertini E.S.^[2], Catteruccia M.^[2]

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P128 - AXIAL MYOPATHIES: WHOLE EXOME SEQUENCING AND GENOTYPE-PHENOTYPE CORRELATION.

Cassandrini D.A.^[2], Vattemi G.^[1], Borin G.U.^[1], Doccini S.^[3], Santorelli F.^[3], Tonin P.^[1]

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P129 - SMPX SPLICING VARIANT EXPANDS THE GENETIC SPECTRUM OF X-LINKED ADULT-ONSET DISTAL MYOPATHY 7

Palmieri F.P.^[1], Zanobio M.^[1], Scarpato M.^[1], Di Letto P.^[1], Romano F.^[1], Giacobbe C.^[2], Picillo E.^[2], Cesarone E.^[1], Ruggiero L.^[3], Nigro V.^[1], Torella A.^[1]

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P130 - HIGH PLASMA NEUROFILAMENT LIGHT CHAIN LEVELS IN INFANTILE-ONSET ASCENDING HEREDITARY SPASTIC PARALYSIS (IAHSP): A PILOT STUDY

Della Vecchia S.^[1], Battaglia N.^[2], Battini R.^[1], Caron G.^[3], Rossi Sebastiano M.^[3], Cesca F.^[4], Ng'Umbi E.E.^[4], Ebrahimi--Fakhari D.^[2], Santorelli F.M.^[1]

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P131 - INVESTIGATING THE MOLECULAR MECHANISMS OF SEPN1-RELATED MYOPATHY USING PATIENT-DERIVED SKELETAL MUSCLE CELLS

Conte E.^[1], Imbriani P.^[1], De Castro F.^[2], Dinoi G.^[1], Lanza M.^[1], Gibertini S.^[3], Maggi L.^[3], Zito E.^[4], De Candia M.^[1], Altomare C.D.^[1], Fanizzi F.P.^[2], De Luca A.^[1], Liantonio A.^[1]

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P132 - UNUSUAL PRESENTATION OF RYR1-RELATED MYOPATHY MIMICKING A METABOLIC MYOPATHY - A CASE REPORT

Virgilio M.^[1], Trucco F.^[3], Brolatti N.^[2], Panicucci C.^[2], Fiorillo C.^[4], Traverso M.^[5], Pedemonte M.^[1], Bruno C.^[2]

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P133 - CONGENITAL MYASTHENIC SYNDROMES (CMS): CLINICAL, GENETIC AND FOLLOW-UP OUTCOMES IN A PEDIATRIC COHORT.

Catteruccia M.^[1], Diodato D.^[1], Tosi M.^[1], Carlesi A.^[2], Fattori F.^[3], Bosco L.^[3], Bertini E.^[1], D'Amico A.^[1]

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P134 - HETEROZYGOUS TTN TRUNCATING VARIANTS IN AXIAL MYOPATHIES: A SIGNIFICANT ENRICHMENT?

Di Feo M.E.^[1], Tasca G.^[2], Töpf A.^[2], Savarese M.^[1], Udd B.^[1], Palmio J.^[3], Jokela M.^[4]

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P135 - EXOME SEQUENCING AND IN SILICO PANEL ANALYSIS AS A DIAGNOSTIC TOOL FOR MUSCLE DISORDERS: HIGH DISCOVERY RATE AND GENETIC HETEROGENEITY IN A PEDIATRIC COHORT

Palombo E.^[1], Giannotta M.^[2], Valentino M.L.^[1], Trentin F.^[3], Scarpini G.^[2], Cordelli D.M.^[3], Carelli V.^[1], Pini A.^[2]

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P136 - QUALITY CONTROL PROGRAM FOR MUSCLE BIOPSIES

Corti S., Comi G., Moggio M., Italian Network F.Q.C.P.

Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico - Italy

P137 - EXPANDING THE PHENOTYPIC SPECTRUM OF MYBPC1-ASSOCIATED CONGENITAL MYOPATHY WITH TREMOR: REPORT OF THE FIRST ITALIAN CASE

Velardo D.^[1], Rimoldi M.^[2], Del Bo R.^[4], Ciscato P.^[1], Croce M.G.^[5], Cosentino G.^[5], Comi G.P.^[2], Corti S.^[1], Ronchi D.^[4], Ravaglia S.^[3]

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P138 - NOVEL ACTN2 VARIANT ASSOCIATED WITH SEVERE CARDIOMYOPATHY WITH PULMONARY HYPERTENSION AND SKELETAL MUSCLE INVOLVEMENT.

Catteruccia M.^{*[1]}, Diodato D.^[1], Magliozzi M.^[2], Adorisio R.^[3], Chiarini Testa M.B.^[4], Verardo M.^[1], D'Amico A.^[1]

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Others P139 - P165

Friday, May 30, 2025 15:45 - 17:30

Chairpersons: *Giovanna Cenacchi (Bologna), Daniele Velardo (Milano)*

P139 - STUDY OF THE MECHANISMS OF ACTION AND NEUROPROTECTIVE EFFECTS OF HEXARELIN AND JMV2894 IN HUMAN SH-SY5Y SOD1G93A CELLS

Meanti R.^[1], Sartori E.^[1], Criscione M.L.^[1], Bresciani E.^[1], Rizzi L.^[1], Fehrentz J.^[2], Denoyelle S.^[2], Torsello A.B.^[1]

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P140 - PRECLINICAL EVALUATION OF THE EFFECTS OF CONJUGATED LINOLEIC ACID ON ANTIOXIDANT ENZYMES GENE EXPRESSION IN A MURINE MODEL OF AMYOTROPHIC LATERAL SCLEROSIS

Claudione L.^[1], Boccanegra B.^[1], De Bellis M.^[1], Bacchetti F.^[2], Milanese M.^[2], Bergamo P.^[3], De Luca A.^[1], Pierno S.^[1]

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P141 - WOMEN HEALTH IN NEUROMUSCULAR DISEASES: TAKING CARE OF NEEDS

Zanolini A.^[1], Ferrari Aggradi C.R.^[1], Colacicco G.^[1], Sacchi V.^[2], Merola V.^[2], Di Raimondo G.^[2], Pisoni M.P.^[2], Sansone V.A.M.^[1]

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P142 - REHABILITATION AND LONG-TERM CARE FOR NEUROMUSCULAR DISORDERS: A CALL TO ACTION

Tabacca A.^[1], Ricci F.^[2], Caterina A.^[3], Rolle E.^[2], Cavallina I.^[2], Oliva M.C.^[1], Ferrante C.^[1], Ruggiero M.^[1], Giannotta G.^[1]

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P143 - ACQUIRED MYOPATHY WITH NEMALINE RODS, CYTOPLASMIC BODIES, RIMMED VACUOLES, AND NECROSIS AND REGENERATION RESPONSIVE TO IVIG: ATYPICAL SLOM, VAMMGAS, IMNM, OR A NEW FORM OF IMMUNE-MEDIATED MYOPATHY?

Vera V., Lauletta A., Merlonghi G., Tufano L., Morino S., Fionda L., Leonardi L., Forcina F., Marando D., Garibaldi M.

Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome - Rome - Italy

P144 - EXPLORING THE CLINICAL AND FUNCTIONAL IMPACT OF TWO CLC-1 CHLORIDE CHANNEL MUTATIONS IN MUSCLE DISEASE

Altamura C.^[1], Laghetti P.^[1], Saltarella I.^[1], Bertoldi S.^[1], Milella G.^[2], Desaphy J.^[1]

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P145 - CLINICAL PATIENT MANAGEMENT SYSTEM (CPMS) IMPLEMENTATION FOR NEUROMUSCULAR PATIENTS' DATA SHARING: "THE ERN EURO-NMD PROJECT"

Fortunato F.^[1], King C.^[1], Neri M.^[1], Gualandi F.^[1], Atalaia A.^[2], Evangelista T.^[3], Ferlini A.^[1]

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P146 - OCCURRENCE OF ASCENDING AORTIC DILATION IN THE FACIO-SCAPULO-HUMERAL DYSTROPHY POPULATION.

Palladino A.^[1], Cesarone E.^[1], D'Ambrosio P.^[2], Salvatore M.^[1], Passamano L.^[1], Scutifero M.^[1], Picillo E.^[1], Politano L.^[2]

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P147 - FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY AND DERMATOMYOSITIS: A CLINICAL CASE REPORT

Di Marco C.^[1], Torchia E.^[1], Ravera B.^[1], Marchese D.^[1], Cacciato E.^[1], Carlomagno V.^[1], Lucchini M.^[2], Bortolani S.^[2], Monforte M.^[2], Mirabella M.^[1], Ricci E.^[1]

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P148 - FSHD ONSET BEFORE 18 YEARS OF AGE: A RETROSPECTIVE LONGITUDINAL STUDY

Ravera B.^[1], Di Marco C.^[1], Barreto-Haagsma A.^[2], Bortolani S.^[3], Torchia E.^[1], Monforte M.^[3], Muni-Lofra R.^[2], Diaz-Manera J.^[2], Guglieri M.^[2], Marini-Bettolo C.^[2], Straub V.^[2], Ricci E.^[1], Tasca G.^[2]

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P149 - REFINING SPORADIC LATE-ONSET NEMALINE MYOPATHY (SLONM): A SINGLE-CENTER EXPERIENCE AND SYSTEMATIC REVIEW OF THE LITERATURE

Lauletta A., Merlonghi G., Forcina F., Fionda L., Leonardi L., Costanzo R., Tufano L., Rossini E., Marando D., Vera V., Antonini G., Morino S., Garibaldi M.

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P150 - ANALYSIS OF FACIAL MUSCLE INVOLVEMENT IN ADULT FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1 PATIENTS THROUGH MAGNETIC RESONANCE AND ULTRASONOGRAPHIC ASSESSMENT

Faedo E.^[3], Pistoia F.^[1], Macciò M.^[2], Susi M.E.^[2], Hamedani M.^[3], Caramellino V.^[4], Massucco S.^[3], Vollaro A.F.M.^[3], Mandich P.^[3], Martinoli C.^[2], Zaottini F.^[2], Grandis M.^[3]

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P151 - EFFICACY OF BISPHTHONATE THERAPY IN PATIENTS WITH NEUROMUSCULAR DISORDERS

Pedemonte M.^[1], Brolatti N.^[2], Panicucci C.^[2], Trucco F.^[1], Casalini S.^[2], Maghnie M.^[3], Striano P.^[1], Di Iorgi N.^[3], Bruno C.^[2]

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P152 - MOTOR REHABILITATION, COMPLEMENTARY ACTIVITIES, AND SPORTS IN CHILDREN WITH NEUROMUSCULAR DISORDERS: AN ANALYSIS OF CARE PATHWAYS AND CORRELATIONS WITH MOTOR SEVERITY

Cavallina I.^[1], Rossi F.^[1], D'Alessandro R.^[1], Rolle E.^[1], Longo L.^[2], Mongini T.E.^[3], Ricci F.S.^[1]

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P153 - SMPX (SMALL MUSCLE PROTEIN X-LINKED) MYOPATHY: A DIAGNOSTIC CHALLENGE IN A RARE DISEASE.

Bertasini M.^[2], Cassandrini D.^[1], Fabrizi G.M.^[2], Santorelli F.M.^[3], Vattemi G.^[2], Tonin P.^[2]

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P154 - EXOBAND® TECHNOLOGY: ENHANCING MOBILITY IN NEUROMUSCULAR DISORDERS THROUGH AN INNOVATIVE PROTOCOL OF MOTOR TESTS AND PROS

Briganti M.^{[1][2]}, Trombetta V.^{[1][2]}, Primiano G.^{[2][3]}, Rossi S.^{[1][3]}, Orlando P.^[1], Robertazzi O.^[1], Servidei S.^{[2][3]}, Sancricca C.^{[1][2]}

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P155 - EARLY ONSET FSHD: AN ITALIAN CASE SERIES

Conti C.^[1], Prestamburgo M.^[1], Iosca A.^[1], Carraro L.^[1], Dainesi I.^[1], Khalil S.^[1], Gardani A.^[2], Maggi L.^[3], D'Alvano G.^[4], Ricci F.S.^[5], D'Alessandro R.^[5], Barp A.^[6], Vercelli L.^[7], Gadaleta G.^[7], Mongini T.E.^[7], Monforte M.^[8], Filosto M.^[9], Ravera B.^[10], Ricci G.^[11], Ruggiero L.^[12], D'Angelo G.^[13], Costantini E.^[14], Verriello L.^[15], Astrea G.^[16], Battini R.^[17], Previtali S.^[18], Pini A.^[19], Fiorillo C.^[20], Berardinelli A.^[2]

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P156 - HOW SPECIFIC ARE "MYOSITIS-SPECIFIC ANTIBODIES"? THE NEED FOR A COMPREHENSIVE APPROACH TO AVOID A MISLEADING INTERPRETATION.

Cicia A., Lucchini M., Carlomagno V., Mirabella M.

Università Cattolica del Sacro Cuore - Rome - Italy

P157 - FOCAL MYOSITIS: A CASE TREATED WITH EXTENSIVE SURGICAL RESECTION

Baratto S.^[1], Panicucci C.^[1], Casabona F.^[1], Gaggero G.^[2], Ricci F.^[3], Brolatti N.^[1], Trucco F.^[4], Pedemonte M.^[4], Fiorillo C.^[4], Consolaro A.^[5], Ravelli A.^[5], Marrè G.^[6], Vellone V.G.^[2], Bruno C.^[1]

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P158 - EXERCISE INTOLERANCE AS EARLY AND MAIN FEATURE IN A PATIENT WITH D4Z4 CONTRACTED ALLELE

Gruosso F.^[1], Majore S.^[2], Colantoni L.^[3], Proietti Piorgio E.^[4], Megalizzi D.^[5], Giardina E.^[5], Petrucci A.^[1]

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P159 - INFANTILE ANTI-HMGCR MYOPATHY: A CASE REPORT

Labella B.^[1], Risi B.^[1], Caria F.^[1], Poli L.^[2], Bozzoni V.^[2], Padovani A.^[2], Filosto M.^[1]

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P160 - ARTEMIS - AN AI-BASED TOOL FOR THE EFFICIENT MORPHOMETRIC ANALYSIS OF HE-STAINED MUSCLE TISSUE SECTIONS.

Markiel M.^[1], Bisceglia G.^[2], Fantini S.^[2], Marczyk M.^[1], Tupler R.^[2], Polanska J.^{*[1]}

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P161 - FIBER DENSITY AS A NEUROPHYSIOLOGICAL MEASURE TO DIFFERENTIATE IBM FROM OTHER INFLAMMATORY MYOPATHIES: PRELIMINARY DATA FROM A PROSPECTIVE STUDY.

Carlomagno V., Cicia A., Di Giovanni J., Tomasello F., Granata G., Lucchini M., Mirabella M.
Catholic University of Sacred Heart - Rome - Italy

P162 - THE ROLE OF THE STABILOMETRIC PLATFORM IN THE ASSESSMENT OF PATIENTS WITH FRIEDREICH'S ATAXIA: AN OBSERVATIONAL STUDY.

Marini V., Dal Molin V., Paparella G., Martinuzzi A.

IRCCS Medea, Department of Conegliano - Conegliano - Italy

P163 - RECURRENT CASE OF ORBITAL EOSINOPHILIC MYOSITIS: CLINICAL INSIGHTS AND CHALLENGES

Ratti A.^[1], Laurini C.^[1], Sorrenti B.^[1], Bosco L.^[1], Yacoub M.^[2], Della Torre E.^[2], Gaiera G.^[3], Criscuoli A.^[4], Filippi M.^[1], Falzone Y.M.^[1], Previtali S.C.^[1]

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P164 - CLINICAL CHARACTERISTICS, TREATMENT PATTERNS, AND POTENTIAL PROGNOSTIC FACTORS IN IDIOPATHIC INFLAMMATORY MYOPATHIES

Bencivenga R.^[1], Spina E.^[2], Ruggiero L.^[3]

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P165 - MYOPATHY IN HIV: VIRAL OR DRUG-INDUCED DAMAGE?

Iaquinta G.^[1], Loprieno S.^[1], Ali G.^[2], Moscato G.^[3], Tagliaferri E.^[4], Siciliano G.^[1], Ricci G.^[1], Torri F.^[1]

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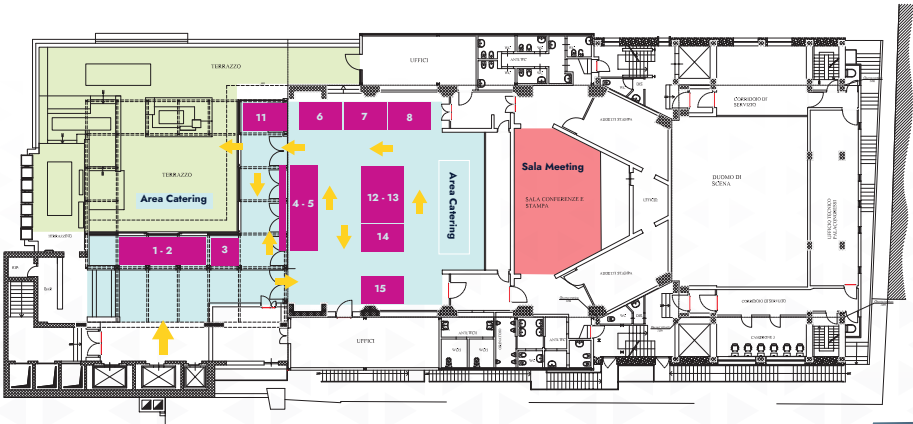
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