

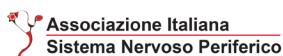


AIM 2025
TAORMINA

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:





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PROGRAMMA

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- 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 2Phenotypes and treatments of TK 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		

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DIRETTIVO**

**COMITATO
SCIENTIFICO**

**COMITATO
ORGANIZZATORE
LOCALE**



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Rosario Rizzuto, <i>Padova</i>	Eric Voorn, <i>Amsterdam</i>
Carmelo Rodolico, <i>Messina</i>	Massimo Domenico Zeviani, <i>Padova</i>
Marcello Romano, <i>Palermo</i>	Ester Zito, <i>Milano</i>
Dario Ronchi, <i>Milano</i>	
Lucia Ruggiero, <i>Napoli</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 DYSTOPHY (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, Biseco 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, Gasperini 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, Pichiechio 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)*

 Onesemonogene 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: *Costanza Lamperti (Milano)*

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, Morrone 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 DINO GMI, 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 I.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, Einvag 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, Ferreira 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.E.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: *Elena Pegoraro (Padova)*

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

¹Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, and Maternal and Child Health (DINO GMI), University of Genoa, Genova - Italy; ²Centro Nacional de Análisis Genómico, Barcelona - Spain; ³John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne - United Kingdom; ⁴Department of Human Genetics, Radboud University Medical Center, Nijmegen - Netherlands; ⁵Centre for Rare Diseases, University Hospital Tübingen, Tübingen - Germany; ⁶Department of Genetics, University Medical Centre Groningen, University of Groningen, Groningen - Netherlands; ⁷Neuroscience and Human Genetics Department, Meyer Children's Hospital IRCSS, Florence - Italy; ⁸Department of Neurology and Epileptology, Hertie Institute for Clinical Brain Research, University of Tuebingen Tuebingen - Germany; ⁹Service de Génétique, Hospices Civils de Lyon, Lyon - France; ¹⁰Myofin Research Group, Folkhalsan Research Center, Helsinki - Finland; ¹¹Montpellier University Hospital, Molecular Diagnostic Laboratory, University Hospital of Montpellier Montpellier - France; ¹²The Dubowitz Neuromuscular Centre, UCL Great Ormond Street, Great Ormond Street Hospital, London - United Kingdom; ¹³Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris - France; ¹⁴Department of Cardiology, Cardiovascular Research Institute Maastricht, Maastricht University Medical Center, Maastricht - Netherlands; ¹⁵Institute for Bioinformatics and Medical Informatics (IBMI), University of Tübingen, Tübingen - Germany; ¹⁶Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Perth - Australia

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

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

²Department of Clinical and Experimental Medicine, University of Pisa, Pisa - Italy; ³NeMO-Brescia Clinical Center for Neuromuscular Diseases, Brescia - Italy; ⁴Department of Clinical and Behavioral Neurology, IRCCS Santa Lucia Foundation, Rome - Italy; ⁵NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Clinical and Experimental Sciences, University of Brescia, Brescia - Italy; ⁶Genomic Medicine Laboratory UILDM, IRCCS Santa Lucia Foundation; Department of Biomedicine and Prevention, Tor Vergata University of Rome, Rome - Italy

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
Altro Domani
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]

[1] Research Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Research Hospital IRCCS, Rome, Italy - Roma - Italy, [2] Center for Experimental Neurotherapeutics, St Jude Children's Research Hospital, Memphis, TN, USA - Memphis - United States of America, [3] MDUK Oxford Neuromuscular

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]

[1]Pediatric Neurology Unit, Catholic University, Rome, Italy - Rome - Italy, [2]Department of Neurosciences, Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy. - Rome - Italy, [3]Section of Genomic Medicine, Department of Life Sciences and Public Health, Catholic University of Sacred Heart, 00168 Rome, Italy. - Rome - Italy, [4]Department of Basic Biotechnological Sciences, Intensive Care and Perioperative Clinics, Catholic University of Sacred Heart, 00168, Rome, Italy. - Rome - Italy, [5]SSD Neuromuscular and Rare Diseases, Department of Neurosciences and Mental Health, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano, Italy - Milano - Italy

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]

[1]Unit of Muscular and Neurodegenerative Disorders, IRCCS Bambino Gesù Children's Hospital - Rome - Italy, [2]Department of General Surgery, Orthopedic Institute, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy, [3]Developmental Neurology Unit, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy

P12 - SCOLIOSIS IN SMA PATIENTS TREATED WITH DISEASE-MODIFYING DRUGS: A PRELIMINAR 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]

[1]Paediatric Neurology, Università Cattolica del Sacro Cuore - Rome - Italy, [2]Nemo Clinical Center, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy, [3]Nemo Clinical Center, Niguarda Hospital - Milan - Italy

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]

[1]Unit of Muscular and Neurodegenerative Disorders, IRCCS Bambino Gesù Children Hospital - Rome - Italy,

[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]

[1]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova - Genova - Italy,

[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]

[1]Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy - Messina - Italy, [2]

Medical Genetics, Department of Precision Medicine, Università degli Studi della Campania 'Luigi Vanvitelli',

Naples, Italy - Napoli - Italy

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]

[1]Neurological Institute, Department of Clinical and Experimental Medicine, University of Pisa, 56126 Pisa, Italy - Pisa - Italy, [2]Unit of Medical Genetics and Neurogenetics, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy - Milan - Italy, [3]Institute of Clinical Human Genetics, University Hospital Regensburg, Regensburg, Germany - Regensburg - Germany, [4]Laboratory of Molecular Genetics, University Hospital of Pisa, Pisa, Italy - Pisa - Italy, [5]Institute of Human Genetics, School of Medicine, Technical University of Munich, Munich, Germany - Munich - Germany

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]}

[1]Mondor Institute of Biomedical Research, University of Paris Est Créteil, INSERM U955, Créteil, France - Créteil - France, [2]AP-HP, North-East Ile-de-France Neuromuscular Pathology Reference Center, Henri Mondor Hospital, Créteil, France [3]Department of Neurosciences, Biomedicine and Movement Science, Section of Clinical Neurology, University of Verona, Verona, Italy [4]Hospital Sant Joan de Déu - Barcelona - Spain, [5]Centre de Référence des Maladies Neuromusculaires NEIdF and ERN EURO-NMD, Neurology Department, University Hospital of Strasbourg - Strasbourg - France, [6]CHU Pasteur - Nice - France, [7]University Hospital Center of Lille, U1172, North-East Ile-de-France Neuromuscular Pathology Reference Center - Lille - France

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]

[1]NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Molecular and Translational Medicine, University of Brescia - Brescia - Italy, [2]Department of Clinical and Experimental Sciences, University of Brescia; Unit of Neurology, ERN EURO-NMD Center "ASST Spedali Civili" - Brescia - Italy, [3]Department of Clinical and Experimental Sciences, University of Brescia; Laboratory of Digital Neurology and Biosensors, University of Brescia - Brescia - Italy, [4]NeMO-Brescia Clinical Center for Neuromuscular Diseases - Brescia - Italy, [5]Unit of Neurology, ERN EURO-NMD Center "ASST Spedali Civili" - Brescia - Italy, [6]Department of Clinical and Experimental Sciences, University of Brescia; Unit of Neurology, ERN EURO-NMD Center "ASST Spedali Civili"; Laboratory of Digital Neurology and Biosensors, University of Brescia - Brescia - Italy, [7]NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Clinical and Experimental Sciences, University of Brescia; Unit of Neurology, ERN EURO-NMD Center "ASST Spedali Civili" - Brescia - Italy

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], Tavian 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]

[1]Amicus Therapeutics Ltd - Milan - Italy, [2]University of Cincinnati College of Medicine - Cincinnati - United States of America, [3]Friedrich-Baur-Institute, Department of Neurology, Ludwig-Maximilians-University - Munich - Germany, [4]Certara GmbH - Lörrach - Germany, [5]Amicus Therapeutics Ltd - Marlow - United Kingdom, [6]Amicus Therapeutics, Inc. - Princeton - United States of America, [7]Amicus Therapeutics GmbH - Munich - Germany, [8]Department of Neurology, University Hospitals Leuven, and Department of Neurosciences, Laboratory for Muscle Diseases and Neuropathies, KU Leuven, Leuven Brain Institute (LBI) - Leuven - Belgium

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]

[1]Unit of Muscular and Neurodegenerative Disorders, Ospedale Pediatrico Bambino Gesù - Roma - Italy, [2]Unit of Gastroenterology and Nutrition, Ospedale Pediatrico Bambino Gesù - Roma - Italy, [3]Unit of Pneumology, Ospedale Pediatrico Bambino Gesù - Roma - Italy

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]

[1]Department of Clinical and Experimental Medicine, University of Messina, Messina - Messina - Italy, [2]Molecular Medicine for Neurodegenerative and Neuromuscular Diseases Unit, IRCCS Stella Maris Foundation, Pisa - Pisa - Italy

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., Iapoco 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]

[1]SC Clinica Neurologica, AOU Sassari. Dipartimento di Medicina, Chirurgia e Farmacia, Università di Sassari - Sassari - Italy, [2]SC Clinica Neurologica, AOU Sassari - Sassari - Italy, [3]Dipartimento di Scienze e Biotecnologie Medico-Chirurgiche, Sapienza Università di Roma - Roma - Italy, [4]Dipartimento di Scienze Biomediche, Università di Sassari - Sassari - Italy

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]

[1]Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome - Rome - Italy, [2]Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital - Rome - Italy

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]

[1]Department of Neurology & Neurological Sciences, Stanford Medicine - Palo Alto - United States of America, [2]argenx - Ghent - Belgium, [3]Harvard Medical School, Massachusetts General Hospital - Boston - United States of America, [4]ZS Associates - Bengaluru - India, [5]ZS Associates - New York - United States of America

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]

[1]Department of Neurology and NeuroCure Clinical Research Center, Charité - Universitätsmedizin Berlin - Berlin - Germany, [2]Neuromuscular and Rare Disease Centre Sant'Andrea Hospital and 3Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), Faculty of Medicine and Psychology, SAPIENZA University of Rome, - Rome - Italy, [3]Stanford Healthcare - Palo Alto - United States of America, [4]Department of Neurology, The University of North Carolina - Chapel Hill - United States of America, [5]Department of Neurology, School of Medicine, International University of Health and Welfare - Tokyo - Japan, [6]argenx - Ghent - Belgium, [7]Ellen & Martin Prosserman Centre for Neuromuscular Diseases, University Health Network and the University of Toronto - Toronto - Canada, [8]Copenhagen Neuromuscular Center, Rigshospitalet, University of Copenhagen, - Copenhagen - Denmark, [9]Department of Neurology, Leiden University Medical Center - Leiden - Netherlands, [10]Department of Neuroimmunology and Neuromuscular Diseases, Fondazione IRCCS Istituto Neurologico "Carlo Besta", - Milan - Italy

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]

[1]Fondazione IRCCS Istituto Neurologico "Carlo Besta", - Milan - Italy, [2]Massachusetts General Hospital - Boston - United States of America, [3]argenx - Ghent - Belgium, [4]Curare Consulting B.V - Liempde - Netherlands, [5]Children's Hospital of Eastern Ontario Research Institute; Division of Neurology, Department of Medicine, The Ottawa Hospital; and Brain and Mind Research Institute, University of Ottawa - Ottawa - Canada

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]

[1]Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy - Milano - Italy, [2]Department of Biomedicine, Neuroscience and Advanced Diagnostics (Bi.N.D.), University of Palermo - Palermo - Italy, [3]Dipartimento di Medicina Clinica e Sperimentale, A.O.U di Messina - Messina - Italy, [4]Dipartimento di Neurologia, Università Cattolica Sacro

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]

[1]Regional Center for Diagnosis and Treatment of Neuromuscular Disease, Department of Biomedicine, Neuroscience and Advanced Diagnostic (BIND), University of Palermo, Palermo, Italy. - Palermo - Italy, [2] Division of Respiratory Medicine, Department PROMISE, "Giaccone" University Hospital, University of Palermo, Palermo, Italy. - Palermo - Italy

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]

[1]Rare Neurological and Neuromuscular diseases center, Neurology Unit, San Filippo Neri Hospital - Rome - Italy, [2]Neurophysiology Unit, Sant'Andrea Hospital - Rome - Italy, [3]NESMOS Department Sapienza University of Rome - Rome - Italy, [4]Neurology Unit, San Filippo Neri Hospital

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], Habetswallner F.^[2], Meacci E.^[4], Marini M.^[2], Habetswallner F.^[1], Iorio R.^[5]

[1]UOC Neurophysiopathology, AORN Cardarelli - Naples - Italy, [2]Department of Neuroscience, Catholic University of the Sacred Heart - Rome - Italy, [3]Neuromuscular and Rare Disease Centre, Sant'Andrea Hospital - Rome - Italy, [4]Department of General Thoracic Surgery, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy, [5]Neurology Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy

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]

[1]UOC Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy; - Milan, Italy - Italy, [2]Clinical Pharmacology and Toxicology School, University of Milan; - Milan - Italy, [3]Primario Emerito Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy - MILAN - Italy

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]

[1]Department of Medicine and Surgery, University of Parma - Parma - Italy, [2]Department of General and Specialized Medicine, University Hospital of Parma - Parma - Italy

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]

[1]Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy; - Milan - Italy, [2]Clinical Pharmacology and Toxicology School, University of Milan; - MILAN - Italy, [3]Primario Emerito, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy; - MILAN - Italy

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]

[1]Neurology Unit, University Hospital "San Giovanni di Dio e Ruggi d'Aragona" - Salerno - Italy, [2]Department of Biomedicine, Neuroscience and Advanced Diagnostic (BIND), University of Palermo - Palermo - Italy, [3]Department of Neuroscience, Imaging and Clinical Sciences, "G. d'Annunzio" University - Chieti - Italy, [4]Department of Neuroscience, University of Salerno, 'Scuola Medica Salernitana' - Salerno - Italy

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]

[1]Catholic University of the Sacred Heart, Rome, Italy - Rome - Italy, [2]Hospital IRCCS Policlinico Agostino Gemelli - Rome - Italy

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]

[1]Department of Biomedicine, Neuroscience, and advanced Diagnostic (BIND), University of Palermo - Palermo - Italy, [2]Neurology Unit, Department of Medicine, Surgery and Dentistry "Scuola Medica Salernitana", University of Salerno - Salerno - Italy

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]

[1]Neuromuscular Unit, Department of Neurosciences "Rita Levi Montalcini", University of Turin - Turin - Italy, [2]Division of Cardiology, Cardiovascular and Thoracic Department, AOU Città della Salute e della Scienza di Torino - Turin - Italy, [3]Department of Neurosciences "Rita Levi Montalcini", University of Turin - Turin - Italy, [4] Department of Neurosciences "Rita Levi Montalcini", AOU Città della Salute e della Scienza di Torino - Turin - Italy, [5]Immunogenetics and Transplant Biology Service, AOU Città della Salute e della Scienza di Torino, University of Turin - Turin - Italy

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]

[1]Department of Medical, Surgical and Experimental Sciences; University of Sassari, Sassari, Italy - Sassari - Italy, [2]Department of Neurosciences, Drugs and Child Health; University of Florence, Florence, Italy - Firenze - Italy

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]

[1]Aldo Moro University of Bari - Bari - Italy, [2]Tor Vergata University - Rome - Italy, [3]Azienda Ospedaliero Universitaria Policlinico di Bari - Bari - Italy, [4]ASL Roma 1 - Roma - Italy, [5]University of Chieti - Chieti - Italy, [6] Center of Innovation in Nursing Research (CINR) - Chieti - Italy

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

Bevilacqua L.^[1], Piscosquito G.^[1], Iovino A.^[1], Toriello A.^[1], Barone P.^[2], Vinciguerra C.^[1]

[1]Neurology Unit, University Hospital "San Giovanni di Dio e Ruggi d'Aragona" - Salerno - Italy, [2]University of Salerno, Department of Medicine Surgery and Dentistry "Scuola Medica Salernitana" Neuroscience Section - Baronissi (SA) - Italy

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]

[1]Department of Clinical and Experimental Medicine, Neurology Unit, University of Pisa - Pisa - Italy, [2] Neurology Unit, Department of Translational Biomedicine and Neurosciences (DiBraiN) - Bari - Italy

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]

[1]Dipartimento Medicina dei Sistemi, Università degli Studi di Roma Tor Vergata - Rome - Italy, [2] Neuromuscular Diseases Unit, Policlinico e Università degli Studi di Roma Tor Vergata - Rome - Italy

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]

[1]Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, University of Turin, Italy - Torino - Italy, [2]Department of Pediatric Pathology and Care "Regina Margherita", Immunology Rheumatology Unit, Children's Hospital Regina Margherita, Turin, Italy - Torino - Italy, [3] Department of Neurosciences RLM, Neuromuscular Unit, University of Turin, Turin, Italy - Torino - Italy

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]

[1]Department of Clinical and Experimental Medicine, Neurology Unit, University of Pisa - Pisa - Italy, [2] Neurology Unit, Departement of Translational Biomedicine and Neurosciences (DiBraiN) - Bari - Italy

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]

[1]Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome - Rome - Italy, [2]Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital - Rome - Italy

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.

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

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]

[1]Unit of Muscular and Neurodegenerative Disorders, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy, [2]Heart Failure, Transplant and Mechanical Cardiorespiratory Support Unit, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy, [3]Department of Anesthesia and Critical Care, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy

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], Veerapandiyan 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]

[1]Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, Rome, Italy - ROMA - Italy, [2]Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health and Institute of Neurology, University College London, & Great Ormond Street Hospital Trust, London, UK - LONDON - U, [3]UC Davis Health, Sacramento, CA, USA - SACRAMENTO - United States of America, [4]Translational Medical Center, National Center of Neurology and Psychiatry, Kodaira, Tokyo, Japan - TOKYO - Japan, [5]Department of Pediatrics, University of Florida, Gainesville, FL, USA - GAINESVILLE - United States of America, [6]Neuromuscular Unit, Neuropaediatrics Department, Hospital Sant Joan de Déu, Fundacion Sant Joan de Déu, CIBERER - ISC III, Barcelona, Spain - BARCELONA - Spain, [7]Children's Hospital of the King's Daughters, Norfolk, VA, USA - NORFOLK - United States of America, [8]Department of Pediatric Neurology, Center for Neuromuscular Disorders in Children and Adolescents, University Clinic Essen, University of Duisburg-Essen, Essen, Germany - ESSEN - Germany, [9]Department of Pediatrics, Division of Neurology, University of Arkansas for Medical Sciences, Arkansas Children's Hospital, Little Rock, AR, USA - ARKANSAS - United States of America, [10]Department of Neurology, Washington University in St Louis, St Louis, MO, USA - ST LOUIS - United States of America, [11]Sarepta Therapeutics, Inc., Cambridge, MA, USA - CAMBRIDGE - United States of America, [12]Roche Products Ltd, Welwyn Garden City, UK - WELWYN GARDEN CITY - United Kingdom, [13]F. Hoffmann-La Roche Ltd, Basel, Switzerland - BASEL - Switzerland

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]

[1]University of California - San Diego - United States of America, [2]Italfarmaco SpA - Milano - Italy, [3]Shriners Hospital for Children - Portland - United States of America, [4]Certara - Princeton - United States of America, [5]CHU Nantes, Reference Centre for Neuromuscular Diseases AOC - Nantes - France, [6]Neuromuscular Research Center, University of California Davis - Sacramento - United States of America, [7]Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS - Rome - Italy

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]

[1]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova, Italy; Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal, and Child Health - DINOEMI, University of Genova - Genova - Italy, [2]Chief of Neurology Division, Nationwide Children's Hospital, Columbus, OH, USA, and Professor of Pediatrics at The Ohio State University College of Medicine, Columbus, OH, USA - Columbus - United States of America, [3]Professor, Neurology and Neurological Sciences (Adult Neurology), of Pediatrics (Genetics) and, by courtesy, of Pathology, Stanford University, Palo Alto, CA, USA - Palo Alto - United States of America, [4]Chair, Department of Physical Medicine & Rehabilitation and Professor, Departments of Pediatrics and Physical Medicine & Rehabilitation, UC Davis, Davis, CA, USA - Davis - United States of America, [5]Pediatric Neurologist and Director of Neurology, Children's Hospital of The King's Daughters, Norfolk, VA, USA - Norfolk - United States of America, [6]Neurologist, UCLA Health, Los Angeles, CA, USA - Los Angeles - United States of America, [7]Professor of Neurology and of Pediatrics, Washington University in Saint Louis, Saint Louis, MO, USA - Saint Louis - United States of America, [8]Sarepta Therapeutics, Inc., Cambridge, MA, USA - Cambridge - United States of America, [9]Roche Products Ltd, Welwyn Garden City, UK - Welwyn Garden City - United Kingdom

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]}

[1]Fondazione UILDM Lazio Onlus, Rome, Italy - Roma - Italy, [2]Department of Medical Surgical Sciences and Translational Medicine, Sapienza University of Rome - Roma - Italy Neurophysiopathology unit, [3]Fondazione Policlinico Universitario A. Gemelli IRCCS - Rome - Italy [4] Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy

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]

[1]University of California - San Diego - United States of America, [2]Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS - Rome - Italy, [3]Italfarmaco S.p.A. - Milan - Italy, [4]Certara - Princeton - United States of America, [5]CHU Nantes, Reference Centre for Neuromuscular Diseases AOC - Nantes - France, [6]Neuromuscular Research Center, University of California Davis - Sacramento - United States of America, [7]Shriners Hospital for Children - Portland - United States of America

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]

[1]University of California Davis Health - Sacramento - United States of America, [2]University of California - San Diego - United States of America, [3]Shriners Hospital for Children - Portland - United States of America, [4]Italfarmaco SpA. - Milan - Italy, [5]Analysis Group, Inc. - Boston - United States of America, [6]CHU Nantes, Reference Centre for Neuromuscular Diseases AOC - Nantes - France, [7]Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS - Rome - Italy

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]

[1]NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Molecular and Translational Medicine, University of Brescia - Brescia - Italy, [2]NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Clinical and Experimental Sciences, University of Brescia - Brescia - Italy, [3]Biochemistry section, Department of Molecular and Translational Medicine, University of Brescia - Brescia - Italy, [4]Radiological Sciences and Public Health, Department of Medical and Surgical Specialties, "Istituto A. Nocivelli" Centre for Research in Molecular Medicine, University of Brescia - Brescia - Italy, [5]Unit of Otolaryngology, ASST "Spedali Civili" - Brescia - Italy, [6]NeMO-Brescia Clinical Center for Neuromuscular Diseases - Brescia - Italy, [7]Unit of Pathology, ASST "Spedali Civili" - Brescia - Italy, [8]Unit of Neurology, ASST "Spedali Civili" - Brescia - Italy, [9]Department of Clinical and Experimental Sciences, University of Brescia; Unit of Neurology, ASST "Spedali Civili" - Brescia - Italy, [10]NeMO-Brescia Clinical Center for Neuromuscular Diseases; Department of Clinical and Experimental Sciences, University of Brescia, Brescia - Brescia - Italy, [11]Medical Genetics Laboratory, Diagnostic Department, ASST "Spedali Civili" - Brescia - Italy

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

Finkel R.^[1], Phan H.C.^[2], Alessi F.^[3], Cazzaniga S.^[3], Bettica P.^[3], Baroni A.^{*,[3]}, Lavery C.G.^[4]

[1]St. Jude Children's Research Hospital - Memphis - United States of America, [2]MD Rare Disease Research - Atlanta - United States of America, [3]Italfarmaco S.p.A. - Milan - Italy, [4]University of California - San Diego - United States of America

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]

[1]School of Medicine and Surgery, University of Milano-Bicocca, Monza, Italy - Monza - Italy, [2]Department of Pharmacy, University of Aldo Moro - Bari - Italy, [3]UMR5247 Institut des Biomolécules Max Mousseron (IBMM) - Montpellier - France

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]

[1]Scientific Institute IRCCS E Medea, Associazione La Nostra Famiglia - Bosisio Parini - Italy, [2]University of California - Davis, Sacramento - United States of America, [3]Santhera Pharmaceuticals (Switzerland) Ltd - Pratteln - Switzerland, [4]John Walton Muscular Dystrophy Research Centre - Newcastle - United Kingdom, [5]Nationwide Children's Hospital - Columbus, Ohio - United States of America, [6]ReveraGen BioPharma - Rockville, Maryland - United States of America, [7]Department of Neurology, University of Pittsburgh School of Medicine - Pittsburgh, Pennsylvania - United States of America

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

Rizzo B.^[1], Dragoni F.^[1], Dainesi M.I.^[2], Di Gerlando R.^[2], Minucchi E.^[2], Berardinelli A.L.^[1], Gagliardi S.^[1]

[1]IRCCS Mondino Foundation - Pavia - Italy, [2]University of Pavia - Pavia - Italy

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]

[1]Pediatric Neurology Unit, Catholic University - Rome - Italy, [2]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, - Genova - Italy, [3]Department of Radiological and Hematological Sciences, Fondazione Policlinico Universitario Agostino Gemelli IRCCS - Rome - Italy, [4]Radiology Unit, IRCCS, Istituto Giannina Gaslini - Genova - Italy, [5]Department of Pediatrics, IRCCS Istituto Giannina Gaslini - Genova - Italy

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]

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

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

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

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]

[1]Translational Cytogenomics Research Unit IRCCS, Bambino Gesù Children's Hospital, Rome - Rome - Italy, [2]Medical Genetics Unit, Department of Medical Sciences, University of Ferrara - Ferrara - Italy, [3]Unit of Muscular and Neurodegenerative Disorders, IRCCS - Rome - Italy, [4]Translational Cytogenomics Research Unit, Laboratory of Medical Genetics, IRCCS - Rome - Italy

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]

[1]Department of Medical Sciences, Unit of Medical Genetics, University of Ferrara - Ferrara - Italy, [2]Department of Medical Sciences, Laboratory for Technologies of Advanced Therapies (LTAT), University of Ferrara - Ferrara - Italy, [3]Department of Drug Science and Technology, University of Turin - Torino - Italy, [4]University College London, Great Ormond Street Hospital for Children and the Francis Crick Institute - London - United Kingdom

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.

Department of Neuroscience, Catholic University Of The Sacred Heart - Rome - Italy

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]

[1]Unit of Muscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS - Rome - Italy, [2]Center of Translational and Experimental Myology, and Dept. of NeuroscienceGenetics, Maternal and Child Health, University of Genova, IRCCS Istituto Giannina Gaslini Rehabilitation Ophthalmology - Genoa - Italy, [3]Pediatric Neurology, Università Cattolica del Sacro Cuore, Rome, Italy Centro Clinico Nemo, Neuropsichiatria Infantile, Fondazione Agostino Gemelli IRCCS - Rome - Italy, [4]Neurology Unit, Fondazione IRCCS Ca Granda Ospedale Maggiore Policlinico, Dino Ferrari Center, Department of Pathophysiology and Transplantation, University of Milan - Milan - Italy, [5]Pediatric Neuromuscular Unit, IRCCS Institute of the , Neurological Sciences of Bologna - Bologna - Italy, [6]Fondazione IRCCS Istituto Neurologico Carlo Besta, Developmental Neurology Unit, - Milan - Italy, [7]The NEMO Center in Milan, Neurorehabilitation Unit, University of Milan, ASST Niguarda Hospital, - Milan - Italy, [8]Institute for Maternal and Child Health, IRCCS,, Burlo Garofolo, - Trieste - Italy, [9]Department of Clinical and Experimental Sciences, University of Brescia (Italy), Brescia, Italy, NeMO-Brescia Clinical Center for Neuromuscular Diseases - Brescia - Italy, [10]Institute of Experimental Neurology (INSPE), Division of Neuroscience, IRCCS San Raffaele Scientific Institute, Milan, ItalyVita-Salute San Raffaele University - Milan - Italy, [11]Medical Genetics and Cardiomyology, University of Campania "Luigi Vanvitelli" - Napoli - Italy, [12]Cardiomyology and Medical Genetics, University of Campania "Luigi Vanvitelli", - Naoli - Italy, [13]Department of Child Neurology and Psychiatry, IRCCS Mondino Foundation - Pavia - Italy, [14]Pediatric Clinic, Department of Medicine and Surgery, University of Parma, - Parma - Italy, [15]Department of Clinical and Experimental Medicine, University of Messina - Messina - Italy, [16]Child Neuropsychiatry Unit, Department of Public Health and Paediatric Sciences, University of Turin, - Turin - Italy, [17]Neurology Unit, Azienda Ospedale Padova Università Padova - Padua - Italy

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]

[1]ChemiCare srl - Novara - Italy, [2]University of Piemonte Orientale - Department of Pharmaceutical Sciences - Novara - Italy, [3]University of Piemonte Orientale - Department of Translational Medicine - Novara - Italy, [4] IRCCS Istituto Giannina Gaslini - Genova - Italy

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]

[1]IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Neuropsichiatria dell'Età Pediatrica, Bologna, Italy UOS Malattie Neuromuscolari dell'Età Evolutiva, IRCCS Istituto delle Scienze Neurologiche di Bologna, Italy. - Bologna - Italy, [2]Department of Medical and Surgical Sciences, University of Bologna, Bologna, Italy - Bologna - Italy, [3]Cardiology Department, Bellaria Hospital, Bologna Italy - Bologna - Italy

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]

[1]Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, University of Turin - Turin - Italy, [2]Department of Mechanical and Aerospace Engineering, Politecnico di Torino - Turin - Italy, [3]Department of Neurosciences RLM, Neuromuscular Unit, University of Turin - Turin - Italy

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]

[1]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini - Genoa - Italy, [2]Core Facilities, IRCCS Istituto Giannina Gaslini - Genoa - Italy

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]

[1]FONDAZIONE POLICLINICO GEMELLI IRCCS - Rome - Italy, [2]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trusts, Newcastle upon Tyne, United Kingdom. - New Castle - United Kingdom

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]

[1]Department of Clinical and Experimental Medicine, University of Pisa - Pisa - Italy, [2]Unità Malattie Metaboliche, Dipartimento di Neuroscienze, Ospedale Meyer Firenze - Firenze - Italy, [3]IRCCS Stella Maris, Calambrone, Pisa - Pisa - Italy, [4]Unit of Epidemiology of Rare Diseases and Congenital Anomalies, Istituto di Fisiologia Clinica - CNR Pisa - Pisa - Italy, [5]Agenzia Regionale di Sanità (ARS) Toscana, Firenze - Firenze - Italy

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]

[1]Department of Biomedical Sciences, University of Padova - Padova - Italy, [2]Department of Comparative Biomedicine and Food Science, University of Padova - Padova - Italy, [3]Department of Biology, University of Padova - Padova - Italy

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]

[1]Foundation IRCCS Neurological Institute Carlo Besta, Neuroimmunology and Neuromuscular Diseases Unit - Milan - Italy, [2]AIDMED, Associazione Italiana Distrofia Muscolare di Emery-Dreifuss - Milan - Italy, [3]patients representatives

P102 - HOMOZYGOUS P.(ILE309SER) VARIANT IN THE SNUPORTIN 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]

[1]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular Unit, Milan, Italy - Milano - Italy, [2]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Medical Genetics Unit, Milan, Italy - Milano - Italy, [3]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milan, Italy, - Milano - Italy, [4]University of Milan, Dino Ferrari Center, Milan, Italy - Milano - Italy

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]

[1]San Camillo IRCCS - Venezia - Italy, [2]Immunology Unit, University Hospital, Azienda Ospedaliera Universitaria Integrata - VERONA - Italy, [3]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular Unit - MILANO - Italy, [4]IRCCS Stella Maris Foundation - PISA - Italy, [5]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit; University of Milan, Dino Ferrari Center - MILANO - Italy, [6]Department of Neurosciences, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona - VERONA - Italy, [7]Department of Medicine, Surgery and Neurosciences, University of Siena - SIENA - Italy, [8]Department of Biomedical and Neuromotor Science, DIBINEM, University of Bologna - BOLOGNA - Italy

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]

[1]Centro Clinico Nemo - Milano - Italy, [2]Department of Health and Sciences, Université du Québec à Chicoutimi, - Chicoutimi - Canada, [3]Memory, Brain & Cognition Lab (UR 7536), University Paris Cité - Paris - France, [4]Queen Square Institute of Neurology, University College London - London - United Kingdom, [5]Regional Pediatric Rehabilitation Center, Queen Silvia Children's Hospital (A-BE) - Gothenburg - Sweden, [6]Department of Pediatric Neurology, Amalia Children's Hospital, Radboud University Medical Center & Donders Institute for Brain, Cognition and Behavior - Nijmegen - Netherlands, [7]Center for Neurological Learning Disabilities, Kempenhaeghe - Heeze - Netherlands, [8]Neurology Department, Centre de Référence de Pathologie Neuromusculaire Paris-Est, Myology Institute, Filière Neuromusculaire, Pitié-Salpêtrière Hospital - Paris - France, [9]Sorbonne Université, Inserm, Centre de Recherche en Myologie, 75013 - Paris - France, [10]University of Florida Genetics Institute, University of Florida - Gainesville - United States of America, [11]Paediatric 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, 16132 -, [12]Department of Neurology, University Hospital Bonn - Bonn - Germany, [13]Maastricht University, Faculty of Health, Medicine and Life Sciences - Maastricht - Netherlands, [14]Maastricht University, Faculty of Health, Medicine and Life Sciences - Maastricht - Netherlands, [15]Department of Neurology, Virginia Commonwealth University - Richmond - United States of America

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]

[1]Nemo Clinical Center, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy, - Roma - Italy, [2]Department of Cardiovascular Medicine, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy - Roma - Italy, [3]. Department of Cardiovascular and Pulmonary Sciences, Catholic University of the Sacred Heart, Largo Agostino Gemelli, Rome, Italy - Roma - Italy, [4]Pediatric Neurology, Università Cattolica del Sacro Cuore, Rome, Italy - Roma - Italy

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]

[1]Università degli Studi di Milano - Milano - Italy, [2]Centro Clinico NeMO Milano - Milano - Italy

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]

[1]Department of Biomedicine and Prevention, Genetics Unit, University of Rome "Tor Vergata", Via Montpellier 1, 00133 Rome, Italy - Rome - Italy, [2]Laboratory of Medical Genetics, Tor Vergata Hospital, Rome, Italy - Rome - Italy, [3]Department of Biotechnology, University of Verona, Verona, Italy - Verona - Italy, [4]Department of Systems Medicine, University of Rome "Tor Vergata", Via Montpellier 1, 00133 Rome, Italy - Rome - Italy

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]

[1]Department of Biomedicine and Prevention, Medical Genetics Section, University of Rome Tor Vergata - Rome - Italy, [2]Laboratory of Medical Genetics, Tor Vergata Hospital - Rome - Italy, [3]Department of Biotechnology, University of Verona - Verona - Italy, [4]Department of Systems Medicine (Neurology), University of Rome Tor Vergata - Rome - Italy

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], Imbrici P.^[2], Cappellari O.^[2], De Luca A.^[2]

[1]Institute of Myology, University of Sorbonne, - Paris - France, [2]Department of Pharmacy & Drug Sciences-Section of Pharmacology, University of Bari Aldo Moro - Bari - Italy, [3]Department of Biosciences Biotechnology and Environment, University of Bari Aldo Moro - Bari - Italy

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]

[1]Department of Biomedicine, Neuroscience, and advanced Diagnostic (BIND), University of Palermo - Palermo - Italy, [2]Oasi Research Institute-IRCCS - Troina - Italy

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.

Neuromuscular Diseases Unit, Department of Systems Medicine, Tor Vergata University of Rome - Rome - Italy

P112 - PREVALENCE OF MENINGIOMA IN MYOTONIC DYSTROPHY TYPE 1

Rossi S.^[4], Dalla Zanna G.^[2], Funcis A.^[2], Sancricca C.^[3], Perna A.^[5], Silvestri G.^[4]

[1]Università Cattolica del Sacro Cuore, Fondazione UILDM Lazio - Rome - Italy, [2]Università Cattolica del Sacro Cuore - Rome - Italy, [3]Fondazione UILDM Lazio, Fondazione Policlinico A. Gemelli IRCCS - Rome - Italy, [4]Fondazione Policlinico A. Gemelli IRCCS, Università Cattolica del Sacro Cuore - Rome - Italy, [5]Ministero della Salute - SASN via Stoppani, 20 - 00054 Fiumicino - Rome - Italy

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.

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

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

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

[1]Department of Clinical and Experimental Medicine, Neurology Unit, University of Pisa - Pisa - Italy, [2]Department of Translational Research and New Technologies in Medicine and Surgery, University of Pisa - Pisa - Italy, [3]O.U. Digestive Endoscopy, University Hospital of Pisa - Pisa - Italy, [4]ENT Audiology and Phoniatrics Unit, University of Pisa - Pisa - Italy, [5]Pulmonary Unit, Cardiothoracic and Vascular Department, Pisa University Hospital - Pisa - Italy

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]

[1]Dipartimento di Scienze Mediche e Chirurgiche (DIMEC), Università di Bologna, Bologna, Italy - Bologna - Italy, [2]IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Neuropsichiatria dell'Età Pediatrica, UOS Malattie Neuromuscolari dell'Età Evolutiva - Bologna - Italy, [3]IRCCS Istituto delle Scienze Neurologiche di Bologna, Programma di Neuroradiologia con Tecniche di elevata complessità - Bologna - Italy, [4]Dipartimento di Scienze Biomediche e Neuromotorie (DIBINEM), Università di Bologna - Bologna - Italy

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]

[1]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular Unit - Milano - Italy, [2]IRCCS C. Mondino Foundation - Pavia - Italy, [3]Department of Neurosciences DNS, University of Padova - Padova - Italy, [4]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit - Milano - Italy, [5] Dino Ferrari Center, University of Milan - Milano - Italy, [6]Department of Clinical and Experimental Medicine, University of Pisa - Pisa - Italy

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]

[1]Department of Neurosciences, DNS, University of Padova - Padova - Italy, [2]Department of Cardiac, Thoracic, and Vascular Science, and Public health, DSCTV, University of Padova - Padova - Italy

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]

[1]Neurology Unit, Department of Clinical and Experimental Medicine, University of Pisa - Pisa - Italy, [2]Laboratory of Molecular Genetics, Azienda Ospedaliero-Universitaria Pisana, Pisa, Italy - Pisa - Italy

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

Liberatoscioli E.^[1], Buldrini B.^[2], Trabanelli C.^[2], Di Muzio A.^[3]

[1]Department of Neuroscience, Imaging and Clinical Sciences, University G. d'Annunzio of Chieti-Pescara, 66100 Chieti, Italy. - Chieti - Italy, [2]Department of Medical Sciences, Medical Genetics Unit, Department of Reproduction and Growth, S. Anna University Hospital, Ferrara, Italy. - Ferrara - Italy, [3]Department of Neurology, "SS Annunziata" Hospital, 66100 Chieti, Italy. - Chieti - Italy

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]

[1]Unit of Neuromuscular Diseases, Department of Systems Medicine, Tor Vergata University of Rome, Via Montpellier 1, 00133 Rome, Italy. - Rome - Italy, [2]Department of Laboratory Medicine, Tor Vergata University Hospital, Viale Oxford 81, 00133 Rome, Italy. - Rome - Italy, [3]Unit of Neuromuscular Diseases, Policlinico Tor Vergata, Rome - Rome - Italy

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]

[1]Pediatric Neurology Unit, Department of Neurosciences, Santobono-Pausilipon Children's Hospital - Naples - Italy, [2]Department of Neurosciences, Reproductive Sciences and Odontostomatology, University of Naples Federico II - Naples - Italy, [3]Medical Genetics, Department of Precision Medicine, Università degli Studi della Campania 'Luigi Vanvitelli' - Naples - Italy

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]

[1]NeuroMuscular Omnicentre (NeMO) Trento, Azienda Provinciale per i Servizi Sanitari (APSS) - Pergine Valsugana (TN) - Italy, [2]Unit of Medical Genetics, Department of Mother and Child, University Hospital S. Anna Ferrara - Ferrara - Italy, [3]Department of Translational Medicine, University of Ferrara - Ferrara - Italy, [4] Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta" - Milano - Italy, [5]DIBINEM - Department of Biomedical and Neuromotor Sciences, Alma Mater Studiorum Università di Bologna - Bologna - Italy, [6]Tecnomed Srl, Centri Diagnostici - Verona - Italy, [7]Laboratory of Medical Genetics, ASST Papa Giovanni XXIII - Bergamo - Italy

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]

[1]Department of Biomedical and Dental Sciences and Morphofunctional Images, University of Messina - Messina - Italy, [2]Molecular Medicine, IRCCS Fondazione Stella Maris - Pisa - Italy, [3]Department of Clinical and Experimental Medicine, University of Messina - Messina - Italy

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]

[1]1 Ospedale Pediatrico Bambino Gesù (OPBG), Roma, Italy - Roma - Italy, [2]2 Centro Nacional de Análisis Genómico (CNAG), Barcelona, Spain - Barcellona - Spain, [3]GENOOX, Amsterdam, Netherlands - Amsterdam - Netherlands, [4]Unit of Medical Genetics, Department of Medical Sciences, Università degli Studi di Ferrara, Ferrara - Ferrara - Italy, [5]GenomeUP, Rome, Italy - Roma - Italy, [6]Roche, Basel, Switzerland - Basel - Switzerland, [7]Illumina, Madrid, Spain - Madrid - Spain, [8]Universitätsklinikum Freiburg, Freiburg, Germany - Freiburg - Germany, [9]CHU Dijon Bourgogne - Dijon - France, [10]Consorzio Futuro in Ricerca - Ferrara - Italy, [11]Sanofi, Brussels, Belgium - Brussels - Belgium

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]

[1]IRCCS Oasi Maria SS - Troina - Italy, [2]University of Palermo - Palermo - Italy

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]

[1]Unit of Muscular and Neurodegenerative Unit, Ospedale Pediatrico Bambino Gesù, [2]Laboratory of Medical Genetics, Ospedale Pediatrico Bambino Gesù

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]

[1]Translational Cytogenomics Research Unit, Bambino Gesù Children Hospital IRCCS - Rome - Italy, [2]Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital IRCCS - Rome - Italy, [3]Developmental Neurology Unit, Bambino Gesù Children's Hospital IRCCS - Rome - Italy, [4]Unit of Neurorehabilitation, Bambino Gesù Children's Hospital IRCCS - Rome - Italy

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]

[1]2 Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona, Verona, Italy - Verona - Italy, [2]1 Immunology Unit, University Hospital, Azienda Ospedaliera Universitaria Integrata, Verona, Italy. - Verona - Italy, [3]3 IRCCS Fondazione Stella Maris, Pisa, Italy - Pisa - Italy

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]

[1]Department of Precision Medicine, University of Campania "Luigi Vanvitelli" - Naples - Italy, [2]Medical Genetics UOSD, Azienda Ospedaliera Universitaria Luigi Vanvitelli - Naples - Italy, [3]Department of Neurosciences, Reproductive and Odontostomatological Sciences, University of Naples "Federico II" - Naples - Italy

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]

[1]IRCCS Fondazione Stella Maris - Pisa - Italy, [2]Movement Disorders Program, Department of Neurology, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA - Boston - United States of America, [3]University of Torino, Molecular Biotechnology and Health Sciences Department, CASSMedChem, 10138 Torino, Italy - Torino - Italy, [4]Department of Life Sciences, University of Trieste, 34127 Trieste, Italy - Trieste - Italy

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]

[1]Department of Pharmacy-Drug Sciences, University of Bari Aldo Moro - Bari - Italy, [2]Department of Biological and Environmental Sciences and Technologies (DiSTeBA), University of Salento - Lecce - Italy, [3] Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta" - Milano - Italy, [4]Istituto di Ricerche Farmacologiche Mario Negri IRCCS - Milano - Italy

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]

[1]Pediatric Neurology and Muscle Disease Unit, IRCCS Istituto Giannina Gaslini - Genova - Italy, [2]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini - Genova - Italy, [3]Pediatric Neurology and Muscle Diseases Unit, IRCCS Istituto Giannina Gaslini and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genoa - Genova - Italy, [4] Child Neuropsychiatry, IRCCS Istituto Giannina Gaslini and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova - Genova - Italy, [5]Unit of Genetics, IRCCS Istituto Giannina Gaslini - Genova - Italy

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]

[1]Unit of Muscular and Neurodegenerative Disorders, Bambino Gesù Children's Research Hospital - Rome - Italy, [2]Developmental Neurology Unit, IRCCS, Bambino Gesù Children's Hospital - Rome - Italy, [3] Translational Cytogenomics Research Unit IRCCS, Bambino Gesù Children's Hospital - Rome - Italy

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]

[1]Folkhälsan Research Centre - Helsinki - Finland, [2]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trusts - Newcastle upon Tyne - United Kingdom, [3]Tampere University Hospital - Tampere - Finland, [4]Division of Clinical Neurosciences, Turku University Hospital and University of Turku - Turku - Finland

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]

[1]IRCCS Istituto delle Scienze Neurologiche di Bologna, Programma di Neurogenetica, Bologna, Italy - Bologna - Italy, [2]IRCCS Istituto delle Scienze Neurologiche di Bologna, Pediatric Neuromuscular Unit, UO Neuropsichiatria dell'Età Pediatrica, Bologna, Italy - Bologna - Italy, [3]Department of Biomedical and Neuromotor Sciences (DIBINEM), University of Bologna, Bologna, Italy - Bologna - Italy

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]

[1]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neuromuscular Unit - Milan - Italy, [2]IRCCS Fondazione Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit - Milan - Italy, [3]IRCCS Fondazione Mondino, Neuromuscular Unit - Pavia - Italy, [4]University of Milan, Dino Ferrari Center - Milan - Italy, [5] Department of Brain and Behavioural Sciences, University of Pavia - Pavia - Italy

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]

[1]Unit of Muscular and Neurodegenerative Disorders, Bambino Gesù Children's Research Hospital - Rome - Italy, [2]Translational Cytogenomics Research Unit RCCS, Bambino Gesù Children's Hospital - Rome - Italy, [3] Heart Failure, Transplant and Mechanical Assist Devices, Bambino Gesù Children's Research Hospital - Rome - Italy, [4]Pediatric Pulmonology and Respiratory Intermediate Care Unit, Sleep and Long Term Ventilation Unit, Bambino Gesù Children's Research Hospital, - Rome - Italy

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]

[1]Università degli Studi di Milano-Bicocca - Monza - Italy, [2]UMR5247 Institut des Biomolécules Max Mousseron (IBMM) - MONTPELLIER - France

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]

[1]University of Bari Aldo Moro - Bari - Italy, [2]University of Genova - Genova - Italy, [3]Institute of biosciences and bioresources, CNR - Napoli - Italy

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]

[1]Centro Clinico Nemo - Milano - Italy, [2]Ostetricia e Ginecologia ASST Grande Ospedale Metropolitano Niguarda - Milano - Italy

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]

[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" - Brindisi - Italy, [2]Child Neuropsychiatric Unit, Department of Public Sciences and Pediatrics, University of Turin - TORINO - Italy, [3]Department of Women's and Children's Health - University of Padua - PADOVA - Italy

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]

[1]Department of Precision and Regenerative Medicine, Section of Pharmacology, School of Medicine, University of Bari Aldo Moro, Bari, Italy - Bari - Italy, [2]Neurology Unit, Department of Basic Medical Sciences, Neurosciences and Sense Organs, University of Bari Aldo Moro, Bari, Italy - Bari - Italy

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]

[1]Unit of Medical Genetics, Department of Medical Sciences, University of Ferrara - Ferrara - Italy, [2]Service of Neuromyology, APHP-GH Pitié-Salpêtrière, Sorbonne Université, - Paris - France, [3]Neuromuscular Morphology Unit, Institute of Myology, GHU Pitié-Salpêtrière, Sorbonne Université - Paris - France

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]

[1]University Hospital of Campania Luigi Vanvitelli; Medical Genetics and Cardiomyology - Naples - Italy, [2]University of Campania Luigi Vanvitelli; Cardiomyology and Medical Genetics - Naples - Italy

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

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

[1]Università Cattolica del Sacro Cuore, Fondazione Policlinico Universitario A. Gemelli IRCSS, Roma, Italy - ROMA - Italy, [2]Unità Operativa Complessa di Neurologia, Fondazione Policlinico Universitario A. Gemelli IRCSS, Roma, Italy - ROMA - Italy

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]

[1]Università Cattolica del Sacro Cuore, Fondazione Policlinico Universitario A. Gemelli IRCCS, Roma, Italy - Roma - Italy, [2]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trusts, Newcastle Upon Tyne, UK. - Newcastle Upon Tyne - United Kingdom, [3]Unità Operativa Complessa di Neurologia, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy - Roma - Italy

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.

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

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]

[1]IRCCS Ospedale Policlinico San Martino - Genoa - Italy, [2]Department of Health Sciences (DISSAL), University of Genoa - Genoa - Italy, [3]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetic and Maternal and Infantile Sciences (DINOEMI), University of Genoa - Genoa - Italy, [4]University of Genoa - Genoa - Italy

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]

[1]Pediatric Neurology and Muscle Diseases Unit, IRCCS Istituto Giannina Gaslini, Genova, Italy - Genova - Italy, [2]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova, Italy - Genova - Italy, [3]Department of Pediatrics, IRCCS Istituto Giannina Gaslini, Genova, Italy - Genova - Italy

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]

[1]Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, University of Turin - Turin - Italy, [2]Unione Genitori Italiani contro il tumore dei bambini (UGI) ODV - Turin - Italy, [3]Department of Neurosciences RLM, Neuromuscular Unit, University of Turin - Turin - Italy

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]

[1]Immunology Unit, University Hospital, Azienda Ospedaliera Universitaria Integrata - Verona - Italy, [2] Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona - Verona - Italy, [3] IRCCS Fondazione Stella Maris - Pisa - Italy

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]}

[1] Fondazione UILDM Lazio Onlus - Rome - Italy [2] Department of Neurosciences, Sensory Organs and Thorax, Agostino Gemelli University Hospital Foundation IRCCS - Rome - Italy [3] Department of Neurosciences, Catholic University of the Sacred Heart - Rome - Italy

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]

[1]Università degli studi di Pavia, Department of Brain and Behavioral Sciences - Pavia - Italy, [2]IRCCS Fondazione Mondino, National Neurological Institute, Department of Child Neurology and Psychiatry - Pavia - Italy, [3]IRCCS Istituto Neurologico Carlo Besta - Milano - Italy, [4]University of Campania "Luigi Vanvitelli" - Napoli - Italy, [5]University of Turin, Child Neuropsychiatric Unit, Department of Public Sciences and Pediatrics - Torino - Italy, [6]Centro Clinico NeMO Trento, Azienda Provinciale per i Servizi Sanitari (APSS) - Pergine Valsunga - Italy, [7]SS Neuromuscular Unit - Department of Clinical and Experimental Sciences, University of Turin - Torino - Italy, [8]Fondazione Policlinico Universitario A. Gemelli IRCCS - Roma - Italy, [9]Department of Clinical and Experimental Sciences, University of Brescia - Brescia - Italy, [10]Università Cattolica del Sacro Cuore - Roma - Italy, [11]Università degli studi di Pisa, Department of Neuroscience - Pisa - Italy, [12]Università degli studi di Napoli Federico II, Dipartimento di neuroscienze e scienze riproduttive ed odontostomatologiche - Napoli - Italy, [13]IRCSS Eugenio Medea- La Nostra Famiglia, Unità Malattie Neuromuscolari - Bosisio Parini - Italy, [14]Centro Clinico NeMo Ancona, Department of Neurology - Ancona - Italy, [15]Azienda sanitaria universitaria Friuli Centrale (ASU FC), Neurology Department - Udine - Italy, [16]Università degli studi di Pisa, Division of Child Neurology and Psychiatry, - Pisa - Italy, [17]IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC di Neuropsichiatria dell'età pediatrica - Bologna - Italy, [18] IRCCS Ospedale San Raffaele, Neurology Department - Milano - Italy, [19]Università degli studi di Bologna, Dipartimento di Scienze Biomediche e Neuromotorie - Bologna - Italy, [20]Università degli studi di Genova, Dipartimento di neuroscienze, riabilitazione, oftalmologia, genetica e scienze materno-infantili - Genova - Italy

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]

[1]Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini - Genoa - Italy, [2] Pathology Unit, IRCCS Istituto Giannina Gaslini - Genoa - Italy, [3]Child Neuropsychiatry Unit, Department of Public Health and Paediatric Sciences, University of Turin - Turin - Italy, [4]Paediatric Neurology and Neuromuscular Disorders Unit, IRCCS Istituto Giannina Gaslini - Genoa - Italy, [5]Paediatric Rheumatology and Autoinflammatory Diseases Unit, IRCCS Istituto Giannina Gaslini - Genoa - Italy, [6]Orthopedic and Traumatology Unit, IRCCS Istituto Giannina Gaslini - Genoa - Italy

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]

[1]Center for Neuromuscular and Neurological Rare Diseases, San Camillo Forlanini Hospital - Rome - Italy, [2]Genetic Unit, San Camillo Forlanini Hospital - Rome - Italy, [3]Genomic Medicine Laboratory-UILDM, Santa Lucia Foundation IRCCS - Rome - Italy, [4]Genomic Medicine Laboratory-UILDM, Santa Lucia Foundation IRCCS - Rome - Italy, [5]Genomic Medicine Laboratory-UILDM, Santa Lucia Foundation IRCCS; Department of Biomedicine and Prevention, Medical Genetics Laboratory, Tor Vergata University - Rome - Italy

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]

[1]NeMO-Brescia Clinical Center for Neuromuscular Diseases, Brescia, Italy - Brescia - Italy, [2]Unit of Neurology, ERN EURO-NMD Center ASST Spedali Civili - Brescia - Italy

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]}

[1]Silesian University of Technology, Department of Data Science and Engineering - Gliwice - Poland, [2] University of Modena and Reggio Emilia, Department of Biomedical, Metabolic and Neural Sciences - Modena - Italy

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]

[1]Neurology Unit, IRCCS San Raffaele Scientific Institute - Milano - Italy, [2]Unit of Immunology, Rheumatology, Allergy, and Rare Diseases (UnIRAR), San Raffaele Scientific Institute - Milano - Italy, [3] Infectious Diseases Department, San Raffaele Scientific Institute - Milano - Italy, [4]Neuro-ophthalmology and Ocular Electrophysiology Center, Department of Ophthalmology, Scientific Institute Capitanio Hospital, Istituto Auxologico Italiano Foundation - Milano - Italy

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

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

[1]AOU San Giovanni e Ruggi d'Aragona, University of Salerno - Salerno - Italy, [2]AORN A. Cardarelli, Napoli - Napoli - Italy, [3]Università Federico II, Napoli - Napoli - Italy

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]

[1]Department of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy - Pisa - Italy, [2]UO Anatomia Patologica, Azienda Ospedaliera Universitaria Pisana, Pisa, Italy - Pisa - Italy, [3]UO Neurologia, Ospedale di Livorno, Livorno, - Livorno - Italy, [4]UO Malattie Infettive, Ospedale di Livorno, Livorno, Italy - Livorno - Italy

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Friday May 30, 2025 12:30-13:30	Giovani AIM	FSHD	Inflammatory Myopathies
Saturday May 31, 2025 08:00-09:00	Myotonic Dystrophies and Myotonias	Rehabilitation and Long-Term Care	Laminopathies Group

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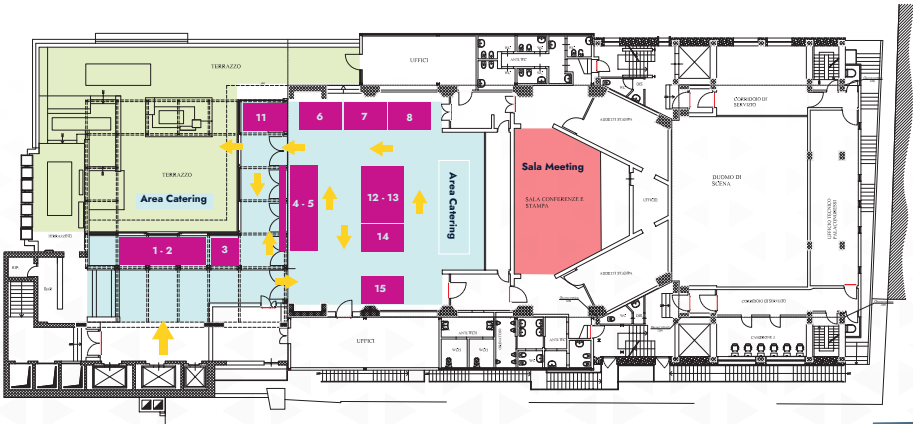
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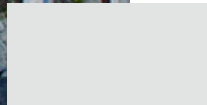


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