



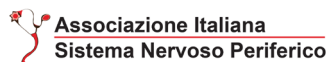
ASSOCIAZIONE ITALIANA
MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

XXIV NATIONAL CONGRESS OF ITALIAN ASSOCIATION OF MYOLOGY

ROME

5 - 8 June 2024

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



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CONSIGLIO DIRETTIVO

Consiglio Direttivo

Prof. Giacomo Pietro Comi - Presidente

Prof. Carlo Minetti - Past President

Prof.ssa Olimpia Musumeci - Segretario

Prof. Luca Bello - Tesoriere

Membri

Dott.ssa Adele D'Amico

Prof. Rocco Liguori

Dott. Lorenzo Maggi

Prof. Michelangelo Mancuso

Prof. Vincenzo Nigro

Dott.ssa Federica Ricci

The background image is a low-angle, upward-looking shot of the interior of the Pantheon in Rome. The image is heavily tinted with a deep blue color. In the foreground, several massive Corinthian columns support a heavy entablature. Above them, a curved balcony or upper level is visible, featuring arched windows and decorative moldings. The top of the image shows the base of the dome's coffered ceiling. Overlaid on this image are several modern graphic elements: in the top left, a large red circle and a white circle overlap, with a thin white line extending from the white circle; in the center right, the word 'FACULTY' is written in white, bold, sans-serif capital letters; below the text, there is a horizontal row of five small circles in red, light blue, white, dark blue, and red; and in the bottom right, a large red circle and a smaller white circle overlap.

FACULTY



FACULTY

Caterina Agosto, Padova
Corrado Angelini, Padova
Giovanni Antonini, Roma
Andrea Barp, Trento
Luca Bello, Padova
Alfredo Berardelli, Roma
Angela Lucia Berardinelli, Pavia
Enrico Bertini, Roma
Silvia Bonanno, Milano
Jim Bonham, Sheffield
Paul Boon, Eindhoven
Annalisa Botta, Roma
Claudio Bruno, Genova
Giorgia Camera, Bergamo
Stefano Canestrari, Bologna
Michela Catteruccia, Roma
Giovanna Cenacchi, Bologna
Giacomo Pietro Comi, Milano
Stefania Corti, Milano
Adele D'Amico, Roma
Maria Grazia D'Angelo, Bosisio Parini
Antonio Di Muzio, Chieti
Vincenzo Di Stefano, Palermo
Daria Diodato, Roma
Carmen Erra, Napoli
Antonio Farina, Lione
Fabiana Fattori, Roma
Alessandra Ferlini, Ferrara
Massimiliano Filosto, Brescia
Chiara Fiorillo, Genova
Laura Fionda, Roma
Matteo Garibaldi, Roma
Francesco Habetswallner, Napoli
Raffaele Iorio, Roma
Maurizio Inghilleri, Roma
Costanza Lamperti, Milano
Rocco Liguori, Bologna
Matteo Lucchini, Roma
Michelangelo Maestri Tassoni, Pisa
Lorenzo Maggi, Milano
Francesca Magri, Milano
Michelangelo Mancuso, Pisa
Roberto Massa, Roma
Riccardo Masson, Milano
Giovanni Meola, Milano
Eugenio Mercuri, Roma
Sonia Messina, Messina
Margherita Milone, Rochester

Carlo Minetti, Genova
Massimiliano Mirabella, Roma
Maurizio Gualtieri Moggio, Milano
Tiziana Mongini, Torino
Mauro Monforte, Roma
Federica Montagnese, Monaco
Olimpia Musumeci, Messina
Vincenzo Nigro, Napoli
Marika Pane, Roma
Giancarlo Parenti, Napoli
Elena Pegoraro, Padova
Elena Pennisi, Roma
Antonio Petrucci, Roma
Daniela Piga, Milano
Antonella Pini, Bologna
Luisa Politano, Napoli
Stefano Previtali, Milano
Guido Primiano, Roma
Susana Quijano Roy, Garches
Sabrina Ravaglia, Milano
Giulia Ricci, Pisa
Enzo Ricci, Roma
Federica Ricci, Torino
Andrea Romigi, Pozzilli
Dario Ronchi, Milano
Carmelo Rodolico, Messina
Vincenzo Russo, Napoli
Francesco Saccà, Napoli
Cristina Sancricca, Roma
Valeria Sansone, Milano
Marco Savarese, Helsinki
Serenella Servidei, Roma
Gabriele Siciliano, Pisa
Gabriella Silvestri, Roma
Antonella Spinazzola, Londra
Maria Sframeli, Messina
Volker Straub, Newcastle upon Tyne
Giorgio Tasca, Newcastle upon Tyne
Paola Tonin, Verona
Yvan Torrente, Milano
Antonio Toscano, Messina
Antonio Trabacca, Brindisi
Federica Trucco, Genova
Antonio Varone, Napoli
Daniele Velardo, Milano
Liliana Vercelli, Torino
Massimo Domenico Zeviani, Padova
Ester Zito, Milano



PROGRAM AT A GLANCE



PROGRAM AT A GLANCE

XXIV National Congress AIM 

Wednesday, 5 June 2024			
	GIOVE HALL	GIUNONE HALL 1	GIUNONE HALL 2
	REGISTRATIONS OPENING		
10:00 - 14:50		POSTER DISPLAY	INTERNATIONAL PRE-CONGRESS COURSE Neuromuscular diagnosis: a multidisciplinary approach from histopathology to molecular genetics. Focus on inflammatory and toxic myopathies.
14:30 - 15:00	Greetings and Introduction		
15:00 - 15:30	MAIN LECTURE I The increasing complexity in the diagnoses and treatment of genetic muscle diseases.		
15:30 - 16:50	WORKSHOP MYOTONIC DYSTROPHY - PART I		
16:50 - 17:10	COFFEE BREAK		
17:10 - 18:10	WORKSHOP MYOTONIC DYSTROPHY - PART II		
18:10 - 19:10	MAIN LECTURE II Preimplantation genetic testing and prenatal diagnosis: fundamental rights and bioethics		
18:40 -19:10	LECTURE - NO CME SPONSORED BY NOVARTIS The latest evidence in the treatment paradigm of Spinal Muscular Atrophy		
20:00	GET TOGETHER COCKTAIL		

PROGRAM AT A GLANCE

XXIV National Congress AIM 

Thursday, 6 June 2024

	GIOVE HALL	GIUNONE HALL 1	GIUNONE HALL 2
08:30 - 09:15	SYMPOSIUM - NO CME SPONSORED BY DYNE Advancements in Neuromuscular Disorder Therapeutics: Lessons Learned in DMD and Emerging Perspectives in DM1 Therapeutic Development	POSTER DISPLAY	
09:45 - 10:15	MAIN LECTURE III Epilepsy in Mitochondrial Disorders		
10:15 - 10:30	COFFEE BREAK		
10:30 - 11:30	Muscle TriviAIM fase eliminatória		
11:30 - 13:00	PLENARY WORKSHOP 1 Current and future perspectives of genomic NBS: are we ready to deal with it?	POSTER SESSION PART I	
12:30 - 14:00	LIGHT LUNCH		
14:00 - 15:00	SYMPOSIUM - NO CME SPONSORED BY BIOGEN Biomarkers and the Development of an Evidence Based Medicine Approach in Spinal Muscular Atrophy		
15:00 - 16:00	SYMPOSIUM - NO CME SPONSORED BY ROCHE THE NEW GENERATION OF SMA PATIENTS		
16.00 - 16.30	COFFEE BREAK		
16:30 - 17:30	ORAL COMMUNICATION MYASTHENIA TRANSVERSAL	ORAL COMMUNICATION MYOTONIC DYSTROPHIE	ORAL COMMUNICATION METABOLIC MYOPATHIES
17:30 - 18:30	ORAL COMMUNICATION MYASTHENIA - THERAPIES	ORAL COMMUNICATION DYSTROPHIES	ORAL COMMUNICATION LATEST DEVELOPMENT IN MYOPATHIES
18.30 - 19.00	MAIN LECTURE IV State of the art of therapy for Duchenne Muscular Dystrophy		
19.00 - 19.30	LECTURE - NO CME SPONSORED BY ITALFARMACO The Italian contribution to scientific innovation in Duchenne musculare dystrophy research		
20.30	SPEAKER DINNER		

PROGRAM AT A GLANCE

XXIV National Congress AIM ●

Friday, 7 June 2024

	GIOVE HALL	GIUNONE HALL 1	GIUNONE HALL 2
08.30 - 10.00	PLENARY WORKSHOP 2 Sporadic Inclusion Body Myositis: Updates on diagnostic clues and therapeutic perspective	POSTER DISPLAY	PARALLEL WORKSHOP 3
10.00 - 10.30	MAIN LECTURE V Optimizing therapeutic options in the management of Pompe disease		
10.30 - 11.00	COFFEE BREAK		
11.00 - 12.30	PLENARY WORKSHOP 4 Mitochondrial diseases: from the emergency department to the new therapies. Avenues and pitfalls	POSTER SESSION PART II	
12.30 - 13.30	LIGHT LUNCH		
13.30 - 14.30	SYMPOSIUM - NO CME SPONSORED BY ARGENX Exploring the role of efgartigimod from clinical trials to real practice in gMG		
14.30 - 15.30	SYMPOSIUM - NO CME SPONSORED BY ALEXION	COFFEE BREAK	
15.30 - 16.00			
16.00 - 17.30	PLENARY WORKSHOP 5 IDIOPATHIC INFLAMMATORY MYOPATHIES: DIAGNOSTIC CHALLENGES, NEW ENTITIES, AND FUTURE THERAPEUTICAL APPROACHES		
17.30 - 18.30	SYMPOSIUM - NO CME SPONSORED BY AMICUS How to improve the management of Pompe Disease		
18.30 - 19.30	General Assembly		
20:30	SOCIAL DINNER		

Saturday, 8 June 2024

	GIOVE HALL	GIUNONE HALL 1	GIUNONE HALL 2
08:30 - 09:30	ORAL COMMUNICATION SMA	ORAL COMMUNICATION FSHD	ORAL COMMUNICATION GENOMIC
09:30 - 10:30	ORAL COMMUNICATION MITOCHONDRIAL	ORAL COMMUNICATION DUCHENNE	ORAL COMMUNICATION SPERIMENTALS
13:30	CLOSING CEREMONY & AWARDS		
13:40	LIGHT LUNCH		



ALF COSTERTIVM FECIT

PROGRAMMA



Wednesday, 5 June 2024

GIOVE HALL

13:00 | REGISTRATION

14:30 - 15:00 | **GREETINGS AND INTRODUCTION**

Alfredo Berardelli (Roma) - Past President SIN

Serenella Servidei (Roma)

Giacomo Pietro Comi (Milano)

15:00 - 15:30 | **MAIN LECTURE I**

Chairperson: *Giacomo Pietro Comi (Milano)*

The increasing complexity in the diagnoses and treatment of genetic muscle diseases.

Volker Straub (Newcastle upon Tyne) - President of WMS

15:30 - 16:50 | **WORKSHOP**

Myotonic Dystrophy - Part I

Chairpersons: *Giovanni Meola (Milano), Gabriele Siciliano (Pisa)*

15:30 | Unravelling the complexity of the DM repeat expansions:
implications for the genotype-phenotype correlations

Annalisa Botta (Roma)

15:45 | Identifying biomarkers of organ involvement in DM1

Gabriella Silvestri (Roma)

16:00 | Towards an Italian consensus on the management of cardiological alterations in the DM

Vincenzo Russo (Napoli)

16:15 | Hypersomnia and sleep-related breathing disorders in myotonic dystrophy type 1
and type 2: diagnosis and treatment

Andrea Romigi (Pozzilli)

16:30 - 16:50 | Discussion

16:50 - 17:10 | Coffee Break

17:10 - 18:10 | **WORKSHOP**

Myotonic Dystrophy- Part II

Chairpersons: *Fabiana Fattori (Roma), Carmelo Rodolico (Messina)*

17:10 | Outcome and intervention in Paediatric DM1

Valeria Sansone (Milano)

17:25 | Treatment development pipeline in the DM1

Roberto Massa (Roma)

17:40 | Myotonic dystrophy type 2: still underdiagnosed? How to improve disease awareness

Federica Montagnese (Monaco)



17:55 - 18:10 | Discussion

18:10 - 18:40 | **MAIN LECTURE II**

Chairperson: *Vincenzo Nigro (Napoli)*

Preimplantation genetic testing and prenatal diagnosis: fundamental rights and bioethics

Stefano Canestrari (Bologna)

18:40 - 19:10 | **LECTURE - SPONSORED BY NOVARTIS**

NO CME

The latest evidence in the treatment paradigm of Spinal Muscular Atrophy.

Chairperson: *Marika Pane (Roma)*

From Clinical Trials to Clinical Practice: anticipating the future and exploring novel aspects of the disease.

Adele D'Amico (Roma), Antonio Varone (Napoli)

20:00 | **GET TOGETHER COCKTAIL**

Thursday, 6 June 2024

GIOVE HALL

08:30 - 09:15 | **SYMPOSIUM - SPONSORED BY DYNE**

NO CME

Advancements in Neuromuscular Disorder Therapeutics:

Lessons Learned in DMD and Emerging Perspectives in DM1 Therapeutic Development

Chairperson: *Eugenio Mercuri (Roma)*

08:30 | Lessons Learned from DMD and Update on the DELIVER Ongoing Phase 1/2 Trial

Marika Pane (Roma)

08:55 | Insights from DM1 Natural History and an Update on the ACHIEVE Ongoing

Phase 1/2 Clinical Trial

Valeria Sansone (Milano)

09:15 - 09:45 | **MAIN LECTURE III**

Chairperson: *Antonio Toscano (Messina)*

Promoting brain health through novel anti-seizure treatments

Paul Boon (Eindhoven) - President European Academy of Neurology

09:45 -10:15 | **MAIN LECTURE IV**

Chairperson: *Antonio Toscano (Messina)*

Epilepsy in Mitochondrial Disorders

Michelangelo Mancuso (Pisa)

10:15 - 10:30 | Coffee Break

10:30 - 11:30 | **MUSCLE TriviAIM- Fase eliminatória**

Chairperson: *Serenella Servidei (Rome), Olimpia Musumeci (Messina)*



11:30 - 13:00 | **PLENARY WORKSHOP 1**

Current and future perspectives of genomic NBS: are we ready to deal with it?

Chairpersons: *Carlo Minetti (Genova), Elena Pegoraro (Padova)*

11:30 | Where are we now

Jim Bonham (Sheffield)

11:45 | Genome NBS: the Screen4Care EU project

Alessandra Ferlini (Ferrara)

12:00 | Barriers and opportunities

Vincenzo Nigro (Napoli)

12:15 | The political side: LEA vs real life

Panel Discussion

12:30 | End of the Session

GIUNONE HALL 1



10:15 - 14:00 | **POSTER SESSION I** **NO CME**

12:30 - 14:00 | Lunch

GIOVE HALL



14:00 - 15:00 | **SYMPOSIUM - SPONSORED BY BIOGEN** **NO CME**

Biomarkers and the Development of an Evidence Based Medicine

Approach in Spinal Muscular Atrophy

Chairperson: *Valeria Sansone (Milano)*

14:00 | Welcome and introduction
Valeria Sansone (Milano)

14:10 | Generating evidence and interpreting data from heterogeneous population
Eugenio Mercuri (Roma)

14:30 | Biomarkers to stratify for disease progression and to monitor treatment efficacy
Stefania Corti (Milano)

14:50 - 15:00 | Discussion

15:00 - 16:00 | **SYMPOSIUM - SPONSORED BY ROCHE** **NO CME**

THE NEW GENERATION OF SMA PATIENTS

Chairperson: *Marika Pane (Roma)*

15:00 | New disease trajectories, new expectations
Marika Pane (Roma)

15:15 | Going beyond motor functions: the bulbar assessment
Riccardo Masson (Milano)

15:30 | Going beyond motor functions: the respiratory assessment
Federica Trucco (Genova)

15:45 - 16:00 | Discussion

16:00 - 16:30 | Coffee Break





16:30 - 17:30 | ORAL COMMUNICATION | MYASTHENIA GRAVIS - TRANSVERSAL

Chairpersons: *Giovanni Antonini (Roma), Rocco Liguori (Bologna)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

16:30 | Ocular myasthenia vs generalized myasthenia gravis with ocular onset:

clinical outcomes from a single-centre retrospective study

ALESSIA PUGLIESE, ADELE BARBACCIA, FIAMMETTA BIASINI, ALBA MIGLIORATO,
CARMEN TERRANOVA, VINCENZO RIZZO, CARMELO RODOLICO
Messina (Italia)

16:42 | Preferential binding to adult or fetal acetylcholine receptor isoform

as a promising predictive biomarker in myasthenia gravis

FRANCESCA BERETTA, EBE SCHIAVO, GREGORIO SPAGNI, SILVIA FALSO,
SARA CORNACCHINI, MASSIMILIANO UGO VERZA, LEONARDO PALAZZO,
LUCA MASSACESI, AMELIA EVOLI, VALENTINA DAMATO
Firenze, Roma (Italia), Berlin (Germany)

16:54 | A multicentre, prospective study comparing autoantibody diagnostic assays in myasthenia gravis

LAURA DEGIGLIO, LAURA CUOMO, MARINA VITILLO, FRANCESCA CORTESE,
MARIANNA BRIENZA, STEFANIA MORINO, LAURA FIONDA, MATTEO GARIBALDI,
GIOVANNI ANTONINI, FRANCESCA GRAGNANI, ANTONIO PETRUCCI,
CARLO PIANTADOSI, MAURIZIO INGHILLERI, MARIA ANTONIETTA INSGRÒ,
MATTEO PRATALI, MARIA TOTARO, ELENA MARIA PENNISI
Roma, Firenze (Italia)

17:06 | Effectiveness of thymectomy in juvenile myasthenia gravis:

preliminary results of a 15-year follow up study

ESTER LATINI, ALBA CEPELE, MELANIA GUIDA, ILARIA CECCARELLI,
CARMELINA CRISTINA ZIRAFÀ, FRANCA MELFI, MICHELANGELO MAESTRI TASSONI
Pisa (Italia)

17:18 | Clinical outcome measures in a prospective cohort of myasthenia gravis patients

MASSIMILIANO UGO VERZA, GREGORIO SPAGNI, SILVIA FALSO, SARA CORNACCHINI,
ELENA CENCINI, LEONARDO PALAZZO, ANTONIO FARINA, ALICE MARIOTTINI,
ALESSANDRO BARILARO, LUCA MASSACESI, AMELIA EVOLI, VALENTINA DAMATO
Firenze, Roma (Italia), Berlin (Germany)



16:30 - 17:30 | ORAL COMMUNICATION | MYOTONIC DYSTROPHIES

Chairpersons: *Costanza Lamperti (Milano), Mauro Monforte (Roma)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

16:30 | Cardiac risk and myocardial fibrosis assessment with Cardiac Magnetic Resonance in patients with Myotonic Dystrophy

ELENA ABATI, VALENTINA TAMBÈ, ANASTASIA ESSERIDOU, GIACOMO PIETRO COMI, STEFANIA CORTI, GIOVANNI MEOLA, FRANCESCO SECCHI
Milano (Italia)

16:42 | Muscle MRI as a biomarker of disease activity and progression in Myotonic Dystrophy type 1: a longitudinal study

LAURA TUFANO, ELISABETTA BUCCI, GIOVANNI ANTONINI, LUCA LEONARDI, MATTEO GARIBALDI
Roma (Italia)

16:54 | Prevalence of AF atrial fibrillation in patients with MD1 Myotonic Dystrophy type 1: a retrospective 16-years follow up

ALESSIA PERNA, FIAMMETTA ALBI, ANTONELLA RISOLI, FEDERICA D'IGNAZIO, CARLA RECUPERO, VITTORIO RISO, LUDOVICO LISPI, ANTONIO PETRUCCI
Roma (Italia)

17:06 | THE METMYD STUDY: baseline data and early results on efficacy and safety of metformin in myotonic dystrophy type 1

ERICA FREZZA, SALVATORE ROSSI, ALESSIA PERNA, ELISABETTA BUCCI, GIULIA GRECO, MARIANGELA GOGLIA, VITTORIO RISO, VIRGINIA VERONICA VISCONTI, ANNALISA BOTTA, MARZIA NUCCETELLI, GIOVANNI ANTONINI, ANTONIO PETRUCCI, GABRIELLA SILVESTRI, ROBERTO MASSA
Roma (Italia)

17:18 | Proof of concept for drug repurposing of fenamates in myotonia congenita

ILARIA SALTARELLA, PAOLA LAGHETTI, CARMEN CAMPANALE, ILARIA NINNI, CONCETTA ALTAMURA, JEAN-FRANCOIS DESAPHY
Bari (Italia)



16:30 - 17:30 | ORAL COMMUNICATION | METABOLIC MYOPATHIES

Chairpersons: *Massimiliano Filosto (Brescia), Elena Pennisi (Roma)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

16:30 | Transition in Glycogen Storage Disease type 2 (GSD2): state of art in Italian Centers

LILIANA VERCELLI, GIULIO GADALETA, GUIDO URBANO, ENRICA ROLLE, SERENA GASPERINI, FRANCESCA MENNI, MICHELE SACCHINI, SABRINA SILIQUINI, ANTONIO TRABACCA, FEDERICA RICCI, TIZIANA MONGINI
Torino, Milano, Ancona, Brindisi (Italia)

16:42 | Quantitative muscle MRI in individual thigh muscles in early Pompe disease

MICHELE GIOVANNI CROCE, LEONARDO BARZAGHI, MATTEO PAOLETTI, CHIARA BONIZZONI, NIELS BERGSLAND, XENI DELIGIANNI, FRANCESCO SANTINI, MASSIMILIANO FILOSTO, BARBARA RISI, LORIS POLI, TIZIANA MONGINI, LILIANA VERCELLI, LORENZO MAGGI, SERENA GASPERINI, MONICA SCIACCO, ANNALISA SECHI, MARINA GRANDIS, MICHELE SACCHINI, SABRINA RAVAGLIA, ANNA PICHIECCHIO
Pavia, Milano, Brescia, Torino, Monza, Udine, Genova (Italia); Basel (Switzerland)

16:54 | Switching treatment to cipaglucosidase alfa plus miglustat positively affects motor function and quality of life in patients with late-onset Pompe disease

ANTONIO TOSCANO, BARRY J. BYRNE, KRISTL G. CLAEYS, PAULA R. CLEMENS, JORDI DÍAZ-MANERA, MAZEN M. DIMACHKIE, PRIYA S. KISHNANI, HANI KUSHLAF, TAHSEEN MOZAFFAR, MARK ROBERTS, NOEMI HUMMEL, MITCHELL GOLDMAN, FRED HOLDBROOK, SIMON SHOHET, BENEDIKT SCHOSER, ON BEHALF OF THE ATB200-07 STUDY GROUP
Messina (Italia); Gainesville, Pittsburgh, Kansas City, Durham, Irvine, Princeton (USA); Leuven (Belgium); Newcastle Upon Tyne, Salford, Marlow (UK); Lorrach, Munich (Germany); Barcelona, Madrid (Spain)

17:06 | Clinical and laboratory follow-up in a cohort of lipid storage myopathies: a single center experience

MATTIA PORCINO, IGNAZIO GIUSEPPE ARENA, ALBA MIGLIORATO, MARIA GRAZIA IGEA FALCONE, CARMELO RODOLICO, ANTONIO TOSCANO, OLIMPIA MUSUMECI
Messina (Italia)

17:18 | A comprehensive evaluation of mobile health technology revealed the ability to identify subtle motor impairment in patients with mild and asymptomatic Pompe disease: one-year follow-up

LUCIA FERULLO, ANDREA RIZZARDI, BEATRICE LABELLA, CINZIA ZATTI, CLINT HANSEN, ROBBIN ROMIJNDERS, BARBARA RISI, FILOMENA CARIA, SIMONA DAMIOLI, EMANUELE OLIVIERI, LORIS POLI, STEFANO COTTI PICCINELLI, WALTER MAETZLER, ALESSANDRO PADOVANI, ANDREA PILOTTO, MASSIMILIANO FILOSTO
Brescia (Italia); Kiel (Germany)



17:30 - 18:30 | ORAL COMMUNICATION | MYASTHENIA GRAVIS - THERAPIES

Chairpersons: Maurizio Inghilleri (Roma), Francesco Saccà (Napoli)

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

17:30 | Achievement of Minimal Symptom Expression in Acetylcholine-Receptor Antibody-Positive Participants with Generalized Myasthenia Gravis and Effect on Disease-Specific Measures in ADAPT/ADAPT+ Studies
 JAMES F. HOWARD, RITA FRANGIAMORE, HIROYUKI MURAI, SRIKANTH MUPPIDI, GLENN PHILIPS, CYNTHIA QI, DEBORAH GELINAS, EDWARD BRAUER, SIHUI ZHAO, VERA BRIL, JOHN VISSING
 North Carolina, Palo Alto (USA); Milano (Italia); Tokyo (Japan); Ontario (Canada); Rigshospitalet (Denmark)

17:42 | Efgartigimod in non-AChR generalized myasthenia gravis
CARLO ANTOZZI, RITA FRANGIAMORE, ELENA RINALDI, FIAMMETTA VANOLI, SILVIA BONANNO, LORENZO MAGGI, FRANCESCA ANDREETTA, ROBERTO ARNABOLDI, ALESSANDRO PINNA, REANTO MANTEGAZZA
 Milano, Roma (Italia)

17:54 | A Real-life experience with Eculizumab and Efgartigimod in generalized Myasthenia Gravis patients
LAURA FIONDA, CHIARA PANE, VINCENZO DI STEFANO, NUNZIA CUOMO, ALESSIO SARNATARO, CLAUDIA VINCIGUERRA, LILIANA BEVILACQUA, FILIPPO BRIGHINA, NICASIO RINI, GIORGIA PUORRO, ANGELA MARSILI, MATTEO GARIBALDI, FRANCESCO SACCÀ
 Roma, Napoli, Palermo, Salerno (Italia)

18:06 | Rituximab in refractory myasthenia gravis: 5 year single center follow up
DARIO RICCIARDI, CARMEN ERRA, FRANCESCO TUCCILLO, FASOLINO ALESSANDRA, BERNARDO MARIA DE MARTINO, FRANCESCO HABETSWALLNER
 Napoli (Italia)

18:18 | Eculizumab in refractory generalized Myasthenia Gravis: a single center 1 year experience
CARMEN ERRA, DARIO RICCIARDI, FRANCESCO TUCCILLO, BERNARDO DE MARTINO, ALESSANDRA FASOLINO, FRANCESCO HABETSWALLNER
 Napoli (Italia)



17:30 - 18:30 | ORAL COMMUNICATION | MUSCULAR DYSTROPHIES

Chairpersons: *Federica Trucco (Genova), Angela Berardinelli (Pavia)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

17:30 | SYNE-1 and SYNE-2 mutations: expanding the genotype and phenotype spectrum of nesprinopathies

MARTA CHELLI, GIULIA MARCHETTO, ANNA RUBEGNI, SARA GIBERTINI, SERENA BARATTO, CLAUDIO BRUNO, MASSIMILIANO FILOSTO, CHIARA FIORILLO, MARINA GRANDIS, DIEGO LOPERGOLO, MARIA ANTONIETTA MAIOLI, ALESSANDRO MALANDRINI, PAOLA MANDICH, ROBERTO MASSA, SABRINA MATÀ, FEDERICO MELANI, LUCIANO MERLINI, ANDREA MIGNARRI, DANIELE ORSUCCI, PATRIZIA SABATELLI, MICHELE SACCHINI, ELISA SCHENA, NILA VOLPI, GIOVANNA LATTANZI, LORENZO MAGGI, FILIPPO MARIA SANTORELLI, PAOLA TONIN, DENISE CASSANDRINI, GAETANO VATTEMI
Verona, Milano, Genova, Brescia, Siena, Roma, Firenze, Bologna (Italia)

17:42 | Emerin is involved in microtubule-organizing center (MTOC) relocalization to the nuclear surface: altered dynamics in type 1 Emery-Dreifuss Muscular Dystrophy

ELISABETTA MATTIOLI, VITTORIA CENNI, ELISA SCHENA CHIARA FIORILLO, ANTONELLA PINI, MELANIA GIANNOTTA, MARCO CAVALLI, ELEONORA CATTIN, FEDERICO CORRADI, ALESSANDRA RECCHIA, GIOVANNA LATTANZI
Bologna, Genova, Modena, Reggio Emilia (Italia)

17:54 | In search of biomarkers for Emery-Dreifuss Muscular Dystrophy

ELEONORA CATTIN, ELISABETTA MATTIOLI, ELISA SCHENA, SILVIA BONANNO, PAOLA CAVALCANTE, STEFANIA MARCUZZO, CLAUDIA MALACARNE, LORENZO MAGGI, MELANIA GIANNOTTA, ANTONELLA PINI, CHIARA FIORILLO, ROBERTA RONCARATI, GAETANO VATTEMI, DENISE CASSANDRINI, GIULIA RICCI, GABRIELE VADI, GABRIELE SICILIANO, ALESSANDRA RECCHIA, GIOVANNA LATTANZI
Reggio Emilia, Modena, Bologna, Milano, Genova, Verona, Pisa (Italia)

18:06 | Novel biomarkers for limb-girdle muscular dystrophy associated to CAPN3 mutation

DIEGO LOPERGOLO, SARA AGUTI, GIAN NICOLA GALLUS, SILVIA BIANCHI, SIMONA SALVATORE, ANNA RUBEGNI, GIANNA BERTI, PATRIZIA FORMICHI, NICOLA DE STEFANO, ALESSANDRO MALANDRINI
Siena, Pisa (Italia)

18:18 | Unraveling of the innate immune system and the complement activation in FSHD cellular muscle models towards the identification of disease biomarkers

ELISABETTA FERRARO, ARIANNA VOTTA, BEATRICE CIURLI, FRANCESCA TORRI, CAROLINA FILIPPONI, CHERYANE LAMA, MICHELE LAI, DAVIDE GABELLINI, EDOARDO MALFATTI, GABRIELE SICILIANO, GIULIA RICCI
Pisa, Milano (Italia); Paris (France)

GIUNONE HALL 2

17:30 - 18:30 | **ORAL COMMUNICATION | LATEST DEVELOPMENT IN MYOPATHIES**

Chairpersons: *Sabrina Ravaglia (Milano), Giulia Ricci (Pisa)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

17:30 | Muscle MRI findings in Italian patients with myofibrillar and distal myopathies: an exploratory analysis from ITA-MeD

SARA BORTOLANI, SILVIA BONANNO, MATTEO LUCCHINI, SABRINA RAVAGLIA, GUIDO PRIMIANO, AMALIA LUPI, MARIA LUCIA VALENTINO, ANTONIO PETRUCCI, MARCO MOSCATELLI, DIEGO LOPERGOLO, MATTEO GARIBALDI, MAURO MONFORTE, CRISTINA SANCRICCA, PIETRO RIGUZZI, LUCA BELLO, ELENA PEGORARO, ELEONORA TORCHIA, ALESSANDRO MALANDRINI, ROCCO LIGUORI, TIZIANA ENRICA MONGINI, MASSIMILIANO MIRABELLA, SERENELLA SERVIDEI, ENZO RICCI, LORENZO MAGGI, GIORGIO TASCA
Roma, Torino, Milano (Italia)

17:45 | Myositis with mitochondrial pathology: a multicentric case series

ANTONIO LAULETTA, LUCA BOSCO, GIOIA MERLONGHI, YURI MATTEO FALZONE, MARTA CHELI, ROBERTA PIERA BENCIVENGA, SARAH LEONARD-LOUIS, OLIVIER BENVENISTE, LUCIA RUGGERO, LORENZO MAGGI, STEFANO PREVITALI, MATTEO GARIBALDI
Roma, Milano, Napoli (Italia); Paris (France)

18:00 | Pathogenic TNNI1 variants cause muscle disease manifesting as either a hypo- (recessively inherited) or a hyper- (dominantly inherited) contractile phenotype

FRANCESCA MAGRI, SIMONA ZANOTTI, DANIELE VELARDO, SARA ANTOGNOZZI, MICHELA RIPOLONE, MOSÈ PARISI, LAURA NAPOLI, PATRIZIA CISCATO, MONICA SCIACCO, STEFANIA CORTI, GIACOMO PIETRO COMI, DARIO RONCHI
Milano (Italia)

18:15 | Treatment of active idiopathic inflammatory myopathies by inhibiting FcRn:

Pre-registration report of ALKIVIA, a phase 2/3 trial with efgartigimod
ROHIT AGGARWAL, MASSIMILIANO MIRABELLA, ANTHONY A. AMATO, DESPOINA PAPADOPOULOU, BAS VAN DER WONING, PAUL DUNCOMBE, INGRID E. LUNDBERG
Pittsburgh, Boston (USA), Roma (Italia), Ghent (Belgium)

GIOVE HALL

18:30 - 19:00 | **MAIN LECTURE V**

Chairperson: *Sonia Messina (Messina)*

State of the art of therapy for Duchenne Muscular Dystrophy
Luca Bello (Padova)

19:00 - 19.30 | **LECTURE - SPONSORED BY ITALFARMACO**

NO CME

The Italian contribution to scientific innovation in research on Duchenne Muscular Dystrophy
Eugenio Mercuri (Roma)



Friday, 7 June 2024

GIOVE HALL

08:30 - 10:00 | **PLENARY WORKSHOP 2**

Sporadic Inclusion Body Myositis: Updates on diagnostic clues and therapeutic perspective

Chairpersons: *Maurizio Moggio (Milano), Antonio Petrucci (Roma)*

08:30 | Latest acquisitions on s-IBM physiopathology and potential impact
for newer therapeutic approaches
Lorenzo Maggi (Milano)

08:55 | The value of the laboratoristic and imaging findings in s-IBM diagnosis and research:
from anti-CN1A antibodies to muscle MRI
Giorgio Tasca (Newcastle upon Tyne)

09:20 | Looking for reliable clinical and instrumental outcome measures in s-IBM
Matteo Lucchini (Roma)

09:45 - 10:00 | Discussion

GIUNONE HALL 2

08:30 - 10:00 | **PARALLEL WORKSHOP 3**

Diagnostic tools for congenital myopathies

Chairpersons: *Enrico Bertini (Roma), Claudio Bruno (Genova)*

08:30 | Clinical framing
Margherita Milone (Rochester)

08:45 | Genomic approach
Marco Savarese (Helsinki)

09:00 | MYO-MRI for congenital myopathies
Susana Quijano Roy (Garches)

09:15 | Biomarkers
Ester Zito (Milano)

09:30 - 10:00 | Discussion

GIOVE HALL

10:00 - 10:30 | **MAIN LECTURE VI**

Chairperson: *Tiziana Mongini (Torino)*

Optimizing therapeutic options in the management of Pompe disease
Olimpia Musumeci (Messina)

10:30 - 11:00 | Coffee Break

GIOVE HALL

11:00 - 12:30 | **PLENARY WORKSHOP 4**

Mitochondrial diseases: from the emergency department to the new therapies. Avenues and pitfalls

Chairpersons: *Antonella Spinazzola (Londra), Dario Ronchi (Milano)*

11:00 | Management of acute metabolic crisis
Serenella Servidei (Roma)

11:18 | Update on Leigh Syndrome
Enrico Bertini (Roma)

11:36 | Therapeutic developments in Friedreich Ataxia
Yvan Torrente (Milano)

11:54 | Focusing on regulatory aspects in neuromuscular disease
Antonella Spinazzola (Londra)

12:12 - 12:30 | Discussion

GIUNONE HALL 1

10:30 - 13:30 | **POSTER SESSION PART II**

NO CME

12:30 - 13:30 | Lunch

GIOVE HALL

13:30 - 14:30 | **SYMPOSIUM - SPONSORED BY ARGENX**

NO CME

Exploring the role of efgartigimod from clinical trials to real practice in gMG

Chairpersons: *Lorenzo Maggi (Milano), Raffaele Iorio (Roma)*

13:30 | Welcoming

13:35 | MG: an IgG mediated disease
Raffaele Iorio (Roma)

13:45 | Long-term efficacy and safety of efgartigimod
Vincenzo Di Stefano (Palermo)

14:00 | Moving beyond clinical trials for patient identification
Lorenzo Maggi (Milano)

14:05 | Real Life experience with efgartigimod
Giorgia Camera (Bergamo)

14:20 - 14:30 | Discussion



14:30 - 15:30 | **SYMPOSIUM - SPONSORED BY ALEXION** **NO CME**
ECULIZUMAB: A YEAR OF VALIDATION FROM TRIAL DATA TO REAL WORLD EXPERIENCE

Chairpersons: *Francesco Habetswallner (Napoli), Roberto Massa (Roma)*

14:30 | Disease Fluctuations Control
Carmen Erra (Napoli)

14:45 | Early Responders vs Late Responders
Silvia Bonanno (Milano)

15:00 | Reduction of Concomitant Therapies
Laura Fionda (Roma)

15:15 - 15:30 | Discussion

15:30 - 16:00 | Coffee Break

16:00 - 17:30 | **PLENARY WORKSHOP 5**

Idiopathic Inflammatory Myopathies:

Diagnostic Challenges, New Entities, And Future Therapeutical Approaches

Chairpersons: *Matteo Garibaldi (Roma), Antonio Di Muzio (Chieti)*

16:00 | New molecular targets for IIMs: from immunopathology to clinical trials
Massimiliano Mirabella (Roma)

16:25 | Immune checkpoint inhibitor-associated myositis:
risk factor, disease courses and therapeutic approach
Antonello Farina (Lione)

16:50 | Muscle MRI and imaging biomarkers: role on diagnosis and follow-up of IIM
Stefano Previtali (Milano)

17:15 - 17:30 | Discussion

GIOVE HALL

17:30 - 18:30 | **SYMPOSIUM - SPONSORED BY AMICUS THERAPEUTICS**

NO CME

How to improve the management of Pompe Disease

Chairs: *Tiziana Mongini (Torino), Antonio Toscano (Messina)*

17:30 | Rationale for the use of small molecules in LSD

Giancarlo Parenti (Napoli)

17:45 | Management and follow-up of the patient with Pompe disease

Cristina Sancricca (Roma)

18:00 | Can cell damage be limited?

Antonio Toscano (Messina)

18:15 - 18:30 | Discussion

18:30 - 19:30 | **GENERAL ASSEMBLY**



08:30 - 09:30 | **ORAL COMMUNICATION | SMA**

Chairpersons: *Caterina Agosto (Padova), Antonella Pini (Bologna)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

08:30 | RAINBOWFISH: Primary efficacy and safety data in risdiplam-treated infants with presymptomatic spinal muscular atrophy (SMA)

RICHARD S FINKEL, MICHELLE A FARRAR, LAURENT SERVAIS, DMITRY VLODAVETS, EDMAR ZANOTELI, MOHAMMED AL-MUHAIZEA, ALEXANDRA PRUFER, LESLIE NELSON, CAROLYN FISCHER, MARIANNE GERBER, KSENIJA GORNI, HEIDEMARIE KLETZ, LAURA PALFREEMAN, ELENI GAKI, PAULO FONTOURA, ENRICO BERTINI, ON BEHALF OF THE RAINBOWFISH STUDY GROUP
Memphis, Dallas (USA); Sydney (Australia); Oxford (UK); Moscow (Russia); Sao Paulo, Rio (Brazil); Riyadh (Saudi Arabia); Basel (Switzerland); Roma (Italia), Liege (Belgium); Paris (France)

08:42 | Early neurological signs in infants identified through neonatal screening for SMA: do they predict outcome?

MARIKA PANE, GIULIA STANCA, CHIARA TICCI, COSTANZA CUTRONA, ROBERTO DE SANCTIS, MATTEO PIRINU, GIORGIA CORATTI, CONCETTA PALERMO, BEATRICE BERTI, DANIELA LEONE, MICHELE SACCHINI, MARGHERITA CERBONESCHI, LAVINIA FANELLI, GIULIA NORCIA, NICOLA FORCINA, ANNA CAPASSO, GIANPAOLO CICALA, LAURA ANTONACI, MARTINA RICCI, MARIA CARMELA PERA, CHIARA BRAVETTI, MARIA ALICE DONATI, ELENA PROCOPIO, EMANUELA ABIUSI, ALESSANDRO VAISFELD, ROBERTA ONESIMO, FRANCESCO DANILO TIZIANO, EUGENIO MERCURI
Roma, Firenze (Italia)

08:54 | Fragility fractures prevalence and bone health in a large cohort of untreated SMA patients

CHIARA PANICUCCI, NOEMI BROLATTI, MARINA PEDEMONTE, FEDERICA RICCI, TIZIANA MONGINI, VALERIA ADA SANSONE, MASSIMILIANO FILOSTO, LUCA BELLO, IRENE BRUNO, VERRIELLO LORENZO, GIULIA RICCI, ADELE D'AMICO, EUGENIO MERCURI, NATASCIA DI IORGI, CLAUDIO BRUNO, ITASMAC GROUP
Torino, Milano, Brescia, Padova, Trieste, Udine, Pisa, Roma (Italia)

09:06 | Single-center long-term follow-up of 18 patients with Spinal Muscular Atrophy (SMA) treated with risdiplam

SIMONA DAMIOLI, BARBARA RISI, FILOMENA CARIA, BEATRICE LABELLA, ENRICA BERTELLA, GIORGIA GIOVANELLI, NESAIBA AIT ALLALI, CHIARA BUCCI, LORIS POLI, ALESSANDRO PADOVANI, MASSIMILIANO FILOSTO
Brescia (Italia)

09:18 | MFM-32 motor scale in adult SMA patients after risdiplam treatment: a single centre experience.

MARIA SFRAMELI, ROBERTO MATERIA, COSIMO ALLEGRA, GIANMARCO BAGNATO, ELENA LA ROSA, SONIA MESSINA
Messina (Italia)

08:30 - 09:30 | ORAL COMMUNICATION | FSHD

Chairpersons: Enzo Ricci (Roma), Corrado Angelini (Padova)

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

08:30 | Enhancing FSHD Diagnosis: a one-year follow-up study on the Efficacy of a Combined Methylation Assay and Machine Learning Pipeline

DOMENICA MEGALIZZI, GIULIA TRASTULLI, EMMA PROIETTI PIORGO, LUCA COLANTONI, RAFFAELLA CASCELLA, CLAUDIA STRAFELLA, ELEONORA TORCHIA, SARA BORTOLANI, MAURO MONFORTE, GUIDO PRIMIANO, CRISTINA SANCRICCA, CARLO CALTAGIRONE, ENZO RICCI, EMILIANO GIARDINA
Roma (Italia); Tirana (Albania)

08:42 | Evaluation of non-invasive biological sources for assessing methylation levels of FSHD-associated locus

CLAUDIA STRAFELLA, DOMENICA MEGALIZZI, GIULIA TRASTULLI, EMMA PROIETTI PIORGO, LUCA COLANTONI, RAFFAELLA CASCELLA, FRANCESCA TORRI, BEATRICE CIURLI, BARBARA RISI, FILOMENA CARIA, SIMONA DAMIOLI, MATTEO GARIBALDI, MARINA GRANDIS, ANTONIO PETRUCCI, CARMELO RODOLICO, ANTONIO DI MUZIO, STEFANO PREVITALI, MAURO MONFORTE, TIZIANA ENRICA MONGINI, LILIANA VERCELLI, VALERIA SANSONE, PAOLA MANDICH, MARIA ANTONIETTA MAIOLI, LUISA POLITANO, MARIANNA SCUTIFERO, ELENA MARIA PENNISI, CARLO CASALI, ENZO RICCI, GUIDO PRIMIANO, CRISTINA SANCRICCA, CARLO CALTAGIRONE, GIULIA RICCI, GABRIELE SICILIANO, MASSIMILIANO FILOSTO, EMILIANO GIARDINA, ITALIAN CLINICAL GROUP FOR FSHD
Roma, Pisa, Brescia, Genova, Messina, Chieti, Milano, Torino, Napoli (Italia); Tirana (Albania)

08:54 | Clinical variability and molecular complexity of FSHD: relevance of deep phenotyping and comprehensive genetics in characterizing atypical clinical cases

FRANCESCA TORRI, CLAUDIA STRAFELLA, LILIANA VERCELLI, GIULIO GADALETA, BARBARA RISI, LUCA COLANTONI, ROSSELLA TUPLER, EMILIANO GIARDINA, MASSIMILIANO FILOSTO, TIZIANA MONGINI, GIULIA RICCI, GABRIELE SICILIANO
Pisa, Roma, Torino, Milano, Reggio Emilia (Italia)

09:06 | Diaphragmatic Ultrasonography in the respiratory assessment of Facioscapulohumeral Muscular Dystrophy (FSHD): a promising approach for management and longitudinal follow-up

ELEONORA TORCHIA, SARA BORTOLANI, BEATRICE RAVERA, RICCARDO INCHINGOLO, ANDREA SMARGIASSI, FRANCESCO MACAGNO, MAURO MONFORTE, GIORGIO TASCA, MATTEO BONINI, LUCA RICHELDI, ENZO RICCI
Roma (Italia), NewCastle Upon Tyne (UK)

09:18 | Molecular imaging of muscle involvement in facioscapulohumeral muscular dystrophy using multispectral optoacoustic tomography

MAURO MONFORTE, SARA BORTOLANI, BEATRICE RAVERA, DAVIDE MARCHESE, YI QIU, ELEONORA TORCHIA, MARCO DE SPIRITO, ENZO RICCI, GIORGIO TASCA
Roma (Italia); Munich (Germany); Newcastle Upon Tyne (UK)

08:30 - 09:30 | ORAL COMMUNICATION | GENOMIC

Chairpersons: Paola Tonin (Verona), Michela Catteruccia (Roma)

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

08:30 | Next-generation sequencing for the diagnosis of primary muscle disorder: the experience of the Dino Ferrari Center in Milan

FRANCESCA MAGRI, DANIELE VELARDO, ELENA ABATI, SARA ANTOGNOZZI, ROBERTO DI BO, DANIELA PIGA, SABRINA LUCCHIARI, SERENA PAGLIARANI, MONICA SCIACCO, STEFANIA CORTI, GIACOMO PIETRO COMI, DARIO RONCHI Milano (Italia)

08:42 | Recurrent TTN missense variants in biallelic titinopathies: a focus on Proline changes

MARIA FRANCESCA DI FEO, MARTIN REES, MATHIAS GAUTEL, HEINZ JUNGBLUTH, MRIDUL JOHARI, KATALIN SZAKSZON, ISTVÁN BALOGH, CLAUDIO BRUNO, CHIARA FIORILLO, MARINA PEDEMONTE, NOEMI BROLATTI, CHIARA PANICUCCI, MONICA TRAVERSO, FRANCESCA FARAVELLI, LUANA MANDARÀ, FRANZISKA SCHNABEL, FEDERICA SILVIA RICCI, ALESSANDRO MUSSA, EDOARDO MALFATTI, ENRICO SILVIO BERTINI, GINA RAVENSCROFT, BJARNE UDD, SAVARESE MARCO Helsinki (Finland); London (UK); Nedlands (Australia); Debrecen (Ungary); Genova, Ragusa, Torino (Italia); Gottingen (Germany); Garches (France)

08:54 | Beyond exome-targeted sequencing approaches to increase the diagnostic rate of unsolved neuromuscular disease patients

PASQUALE DI LETTO, ANNALaura TORELLA, SARAH IFFAT RAHMAN, MARIA ELENA ONORE, ESTHER PICILLO, GIULIO PILUSO, VINCENZO NIGRO Napoli (Italia)

09:06 | Computational Models for new Patients Stratification Strategies of Neuromuscular Disorders: a new strategy to tackle hereditary neuromuscular disorders

BENEDIKT SCHOSER, JOANNA POLANSKA, VOLKER STRAUB, MARCO SVARESE, FILIPPO SANTORELLI, JOCELYN LAPORTE, MARCELLO SCIPIONI, MICHAEL OBACH, PETER STEENSGAARD, ANNALISA DE ANGELIS, ROSSELLA TUPLER München, Tübingen (Germany); Gliwice (Poland); Newcastle Upon Tyne (UK); Helsinki (Finland); Pisa, Roma, Modena (Italia); Illkirch (France); Lugano (Switzerland); San Sebastian (Spain)

09:18 | Beyond the exome: a very complex diagnosis of myofibrillar myopathy solved by genomic long read sequencing

ANNALaura TORELLA, GIULIO PILUSO, MARIA ELENA ONORE, PASQUALE PRIMO, PASQUALE DI LETTO, MARIATERESA ZANOBIO, CARMINE SPAMPANATO, GERMAN DEMIDOV, VIVIANA CETRANGOLO, SARAH IFFAT RAHMAN, FRANCESCA TORRI, GIULIA RICCI, GABRIELE SICILIANO, VINCENZO NIGRO Napoli, Pisa (Italia); Tübingen (Germany)



09:30 - 10:30 | ORAL COMMUNICATION | MITOCHONDRIAL

Chairpersons: *Daria Diodato (Roma), Massimo Zeviani (Padova)*

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

09:30 | Developing AAV gene therapy for mitochondrial myopathies

VALERIA BALMACEDA, RAFFAELE CERUTTI, MASSIMO ZEVIANI, CARLO VISCOMI
Padova (Italia)

09:42 | Urolithin A as a potential treatment for mitochondrial myopathies

VALERIA BALMACEDA, RAFFAELE CERUTTI, SARA VOLTA, MASSIMO ZEVIANI,
CARLO VISCOMI
Padova (Italia)

09:54 | A Pilot Phase 2 Randomized Trial to Evaluate the Safety and Potential Efficacy of Etravirine in Friedreich Ataxia Patients

ANDREA MARTINUZZI, GABRIELLA PAPARELLA, CRISTINA STRAGÀ, NICOLA PESENTI,
ROBERTO TESTI
Roma (Italia)

10:06 | Serum neurofilament light chain in mitochondrial diseases: exploring a new promising biomarker.

GUIDO PRIMIANO, ANGELA ROMANO, MARCO LUIGETTI, ANDREA SABINO,
MICHELANGELO MANCUSO, PIERVITO LOPRIORE, VINCENZO MONTANO,
VALERIO CARELLI, CHIARA LA MORGIA, MARIA LUCIA VALENTINO,
COSTANZA LAMPERTI, VALERIA NICOLETTA, ALESSIA CATANIA, MASSIMILIANO
FILOSTO, BARBARA RISI, CRISTINA SANCRICCA, DOMENICO PLANTONE,
SERENELLA SERVIDEI
Roma, Pisa, Bologna, Milano, Brescia, Siena (Italia)

10:18 | Role of muscle biopsy in mitochondrial myopathy: genotype-phenotype correlation

DARIO ZOPPI, RUGGIERO DI LEO, ANNA RUSSO, ROBERTA PIERA BENCIVENGA,
FLORIANA VITALE, ROSARIO RUSSO, VIRGINIA BOEMIA, DENISE CASSANDRINI,
CLAUDIA NESTI, FILIPPO MARIA SANTORELLI, LUCIA RUGGIERO
Napoli, Pisa (Italia)

09:30 - 10:30 | ORAL COMMUNICATION | DUCHENNE MUSCULAR DYSTROPHY

Chairpersons: Luisa Politano (Napoli), Stefano Previtali (Milano)

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

09:30 | Safety and efficacy of delandistrogene moxeparvovec versus placebo in Duchenne muscular dystrophy (EMBARK): Pivotal Phase 3 Primary results

JERRY R. MENDELL, FRANCESCO MUNTONI, CRAIG M. MCDONALD, EUGENIO MERCURI, EMMA CIAFALONI, HIROFUMI KOMAKI, CARMEN LEON-ASTUDILLO, ANDRÉS NASCIMENTO, CRYSTAL PROUD, ULRIKE SCHARA-SCHMIDT, ARAVINDHAN VEERAPANDIYAN, CRAIG M. ZAIDMAN, MAITEA GURIDI, ALEXANDER P. MURPHY, CAROL REID, CHRISTOPH WANDEL, DAMON R. ASHER, EDDIE DARTON, STEFANIE MASON, RACHAEL A. POTTER, TEJI SINGH, WENFEI ZHANG, PAULO FONTOURA, JACOB S. ELKINS, LOUISE R. RODINO-KLAPAC, ON BEHALF OF THE EMBARK STUDY GROUP
Columbus, Sacramento, Rochester, Gainesville, Norfolk, Washington, Cambridge (USA); London, Welwyn Garden City (UK); Roma (Italia); Tokyo (Japan); Essen (Germany); Basel (Switzerland)

09:42 | Extensive next-generation sequencing approaches for the identification of rare genomic structural variants involving the DMD gene

ALICE MARGUTTI, MARTINA MIETTO, VITTORIA NAGLIATI, MARIA SOFIA FALZARANO, GAETANO DE FEO, GIUSY CAVARRETTA, FERNANDA FORTUNATO, MARCELLA NERI, FRANCESCA GUALANDI, ALESSANDRA FERLINI, RITA SELVATICI
Ferrara (Italia)

09:54 | Gut microbiota signature in Duchenne Muscular Dystrophy

CHIARA PANICUCCI, SARA CASAINI, GIOVANNI FIORITO, ALESSANDRA BIOLCATI RINALDI, DAVIDE CANGELOSI, AGNESE REPETTO, NOEMI BROLATTI, MARINA PEDEMONTE, ELISA PRINCIPI, ANTONELLA RIVA, CRISTINA VENTURINO, PASQUALE STRIANO, PAOLO UVA, CLAUDIO BRUNO
Genova (Italia)

10:06 | The hidden face of Duchenne (Neuro)Muscular Dystrophy: social cognition impairment as a feature of the neuropsychological phenotype of DMD

STEFANO PARRAVICINI, CARLO ALBERTO QUARANTA, MARIA IRENE DAINESI, SELVIA KHALIL, ANGELA BERARDINELLI
Pavia (Italia)

10:18 | Nonsense mutations in Becker muscular dystrophy:

DMD gene position and disease severity

DOMENICO GORGOLIONE, DANIELE SABBATINI, PIETRO RIGUZZI, GIULIANA CAPECE, MARIKA PANE, SERENELLA SERVIDEI, MARTA BRIGANTI, CRISTINA SANCRICCA, FABIO BRUSCHI, ANNA ARDISSONE, RICCARDO MASSON, ANNAMARIA GALLONE, LORENZO MAGGI, ESTHER PICILLO, ANGELA PETROSINO, SARA VIANELLO, MARTINA PENZO, MATTEO VILLA, MARIA SFRAMELI, COSIMO ALLEGRI, ANDREA BARP, ALESSANDRA DI BARI, FRANCESCA SALMIN, EMILIO ALBAMONTE, GIOVANNI COLACICCO, CHIARA PANICUCCI, MONICA TRAVERSO, CONCETTA PALERMO, ALBERTO LERARIO, DANIELE VELARDO,

MARIA GRAZIA D'ANGELO, ANGELA BERARDINELLI, ALICE GARDANI,
ROBERTA NICOTRA, STEFANO PARRAVICINI, GABRIELE SICILIANO, GIULIA RICCI,
FRANCESCA TORRI, GIULIO GADALETA, GUIDO URBANO, ENRICA ROLLE,
FEDERICA RICCI, ADELE D'AMICO, MICHELA CATTERUCCIA, ANTONELLA PINI,
MELANIA GIANNOTTA, ROBERTA BATTINI, GEMMA MARINELLA,
STEFANO CARLO PREVITALI, ALBERTO ANDREA ZAMBON, ALESSANDRA FERLINI,
FERNANDA FORTUNATO, FRANCESCA MAGRI, TIZIANA MONGINI,
VALERIA ADA SANSONE, CLAUDIO BRUNO, SONIA MESSINA, VINCENZO NIGRO,
LUISA POLITANO, ISABELLA MORONI, EUGENIO MERCURI, LUCA BELLO,
ELENA PEGORARO
Padova, Roma, Milno, Napoli, Messina, Genova Bosisio Parini, Pavia, Torino, Pisa,
Bologna, Ferrara (Italia)

GIUNONE HALL 2

09:30 - 10:30 | ORAL COMMUNICATION | EXPERIMENTAL MODELS

Chairpersons: Daniela Piga (Milano), Giovanna Cenacchi (Bologna)

The Presenting Author has 10 min. for the presentation and 2 min. for the discussion

09:30 | Creation of a Zebrafish model of LGMDD2 and role of TNPO3 in the pathogenetic mechanism

MARIA TERESA RODIA, ROBERTA COSTA, MARTINA FAZZINA, MATTEO BERGONZONI,
FRANCESCO BORGIA, RAFFAELLA CASADEI, GIUSEPPE SABBIONI,
MARIA TERESA ALTIERI, GIULIA BREVEGLIERI, MONICA BORGATTI, ELIA GATTO,
CRISTIANO BERTOLUCCI, GIOVANNA CENACCHI, FLAVIA FRABETTI
Bologna, Ferrara (Italia)

09:42 | Yeast NDI1 rescues muscular NDUFS3-/- mouse model: a new prospect of flexible gene therapy for complex-I deficiencies

DANIELE SALA, SILVIA MARCHET, COSTANZA LAMPERTI, HOLGER PROKISCH,
NATHALIA DRAGANO, BIRGIT RATHKOLB, ANTONIO AGUILAR-PIMENTEL,
OANA AMARIE, LORE BECKER, JULIA CALZADA-WACK, PATRICIA DA SILVA-BUTTKUS,
LILLIAN GARRETT, SABINE HÖLTER, ADRIÁN SANZ-MORENO, NADINE SPIELMANN,
HELMUT FUCHS, VALERIE GAILUS-DURNER, MARTIN HRABĚ DE ANGELIS
Milano (Italia); Neuherberg, Munich (Germany)

09:54 | Engineering 3D Models to Investigate LGMDD2 Transportin 3 Related: Insights into Myogenic Processes and Contractile Dysfunction

SERAFINA PACILIO, ROBERTA COSTA, MARIA TERESA RODIA, SARA LOMBARDI,
LUANA DI LISA, GHISLAIN BANOS, NATHALIE DIDER, EDOARDO MALFATTI,
GIOVANNA CENACCHI, MARIA LETIZIA FOCARETE
Bologna (Italia); Creteil (FRance)

10:06 | Modeling Myotonic Dystrophy type 2 in human cerebral organoids

PAOLA SPITALIERI, ELISA MACRÌ, FEDERICA CENTOFANTI, VIRGINIA VERONICA
VISCONTI, MARIA GIOVANNA SCIOLI, ANNALUCIA SERAFINO, MARZIA ROSSATO,
MARCO CARLOMAGNO, ROSANNA CARDANI, GIUSEPPE NOVELLI, FEDERICA
SANGIUOLO, ANNALISA BOTTA
Roma, Verona, Milano (Italia)

10:18 | Patient-derived organoids as a new in vitro paradigm to study Myotonic Dystrophy type 1
LORENZO FONTANELLI, AMANDA HUANG, ERIKA SCHIRINZI, GIULIA RICCI, AITOR AGUIRRE, GABRIELE SICLIANO
Pisa (Italia)

10:30 - 11:30 | MUSCLE TriviAIM - THE FINAL
Chairpersons: *Serenella Servidei (Rome), Giacomo Pietro Comi (Milan)*



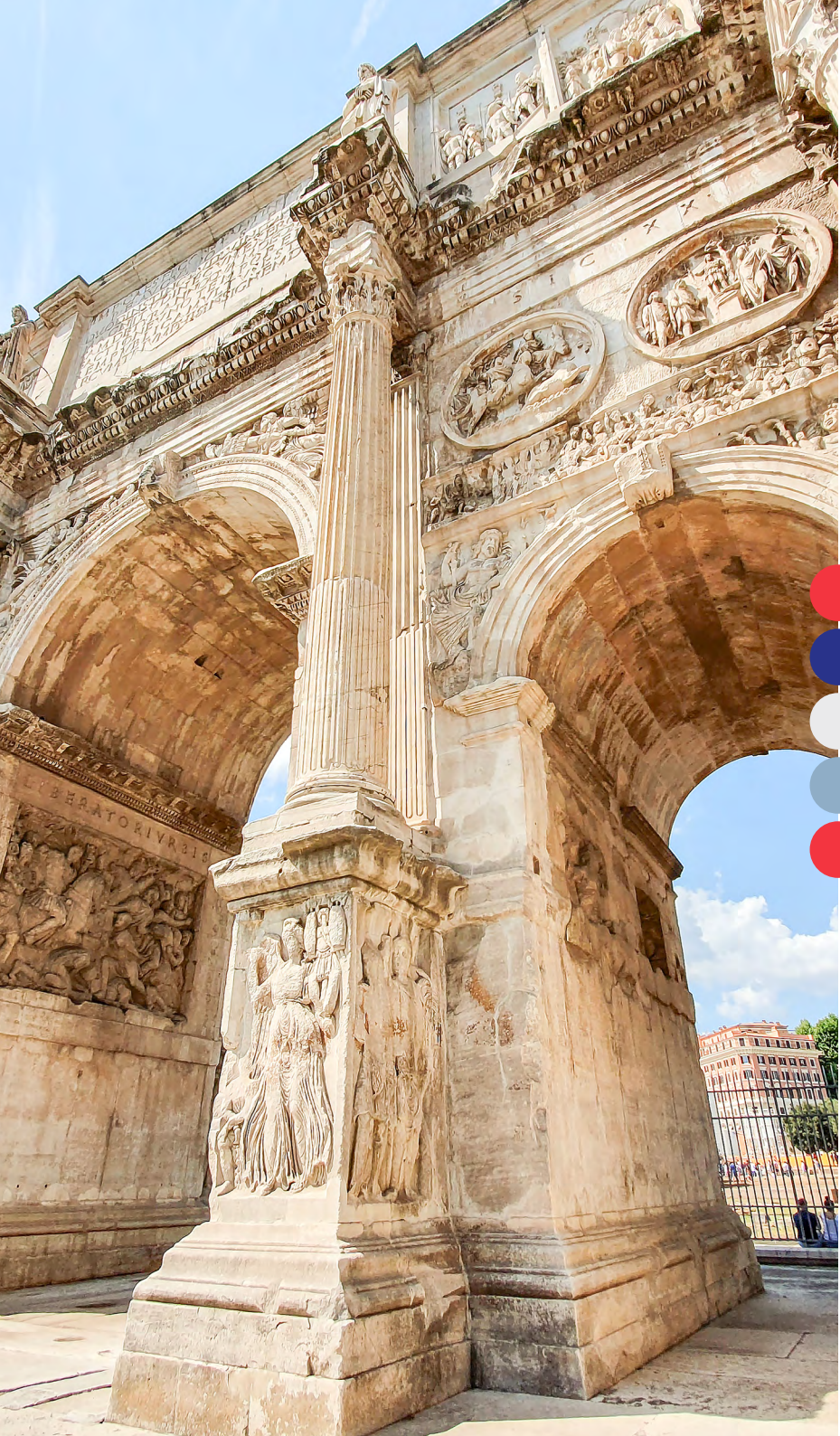
11:30 - 13:30 | ROUND TABLE MEETING with PATIENT'S ASSOCIATIONS
Chairperson: *Giacomo Pietro Comi (Milano), Federica Ricci (Torino)*

Intervengono:

<i>Francesco Ieva</i>	AltroDomani e Consulta Malattie Neuromuscolari
<i>Elisabetta Conti</i>	AIG 
<i>Daniela Lauro</i>	Famiglie SMA 
<i>Beatrice Vola</i>	GFB Gruppo Familiari Beta-sarcoglicanopatie
<i>Paula Morandi</i>	Mitocon
<i>Fernanda De Angelis</i>	Parent Project
<i>Danila Baldessarri</i>	Telethon
<i>Stefania Pedroni</i>	UILDM

13:30 | AWARDS AND CONCLUSIONS

13:40 | Light Lunch





POSTER SESSION



Poster Session

Spinal Muscular Atrophies and Moto-Neuron Disorders

Thursday June 6, 2024 10:15 - 14:00

Chairperson: *Daniele Velardo (Milano)*

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P01 - Targeting upper motor neurons: a neural stem cell treatment for Amyotrophic Lateral Sclerosis

LORENZO QUETTI¹, LUCA SALI¹, MATILDE CONTARDO², ROBERTA DE GIOIA¹, MONICA NIZZARDO¹, ALBERTO ROMANO¹, LORENZO BRAMBILLA¹, LINDA OTTOBONI², FEDERICA RIZZO², STEFANIA CORTI^{1,2}, GIACOMO PIETRO COMI^{1,2}
1. Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy;
2. Università degli Studi di Milano, Dep. Pathophysiology and Transplantation, Milan, Italy

P02 - Basal metabolism, myokine levels and disease severity in Amyotrophic Lateral Sclerosis

MARIANGELA GOGLIA¹, LAURA BOFFA¹, ERICA FREZZA¹, GIULIA GRECO¹, FRANCESCO GRUOSSO¹, ILARIA PETITTA¹, GIULIA NARDINO¹, GIOVANNI VIETRI¹, MARZIA NUCCETELLI², ANTONINO DE LORENZO³, ROBERTO MASSA¹
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P03 - Exploring the effects of Risdiplam-like compound on Spinal Muscular Atrophy using a 3D stem cell-derived spinal cord model

FRANCESCA BEATRICE¹, ANDREA D'ANGELO¹, JESSICA ONGARO², PAOLA RINCHETTI¹, IRENE FARAVELLI¹, MATTEO MIOTTO³, SIMONA LODATO³, GIACOMO COMI^{1,2}, LINDA OTTOBONI¹, STEFANIA CORTI^{1,2}
1. University of Milan, Dept. of Pathophysiology a Transplantation, Milan, Italy; 2. Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; 3. Humanitas University, Milan, Italy

P04 - Need for tube feeding in SMA type I patients treated with disease modifying therapies: prognostic factors

Presented by CHIARA ARPAIA
LAURA ANTONACI¹, MARIKA PANE¹, GIULIA STANCA¹, GIORGIA CORATTI¹, ADELE D'AMICO², VALERIA SANSONE³, BEATRICE BERTI¹, LAVINIA FANELLI¹, EMILIO ALBAMONTE¹, CAROLINA AUSILI¹, ANTONELLA CERCHIARI², MICHELA CATTERUCCIA², ROBERTO DE SANCTIS¹, DANIELA LEONE¹, CONCETTA PALERMO¹, BIANCA BUCHIGNANI¹, ROBERTA ONESIMO¹, MICHELE TOSI², MARIA CARMELA PERA¹, CHIARA BRAVETTI¹, ENRICO BERTINI², EUGENIO MERCURI¹
1. Fondazione Policlinico Gemelli IRCSS; 2. Ospedale Pediatrico Bambino Gesù Irccs; 3. Centro Clinico Nemo - Rome, Italy

P05 - Assessment of motor endurance in spinal muscular atrophy (EnduSMA protocol): one-year follow-up data of a multicenter cohort study

ROBERTO CHIAPPINI¹, GIULIA RICCI¹, FRANCESCA TORRI¹, ALESSANDRA GOVONI¹, LAURA MANCA², GABRIELE VADI¹, STEFANO ROCCELLA³, FRANCESCA MAGRI⁴, MEGI MENERI⁵, FEDERICA FASSINI⁴, VERIA VACCHIANO⁶, SILVIA TOMASSINI⁵, NOEMI GIRONELLA⁵, MICHELA COCCIA⁵, GIACOMO COMI⁴, ROCCO LIGUORI⁶, GABRIELE SICILIANO¹
1. Neurology Unit, Department of Clinical And Experimental Medicine, University of Pisa, Pisa,

Italy; 2. Department of Mathematics, University of Pisa, Pisa, Italy; 3. The Biorobotics Institute, Sant'anna Superior Studies School, Pisa, Italy
4. Neurology Unit, Fondazione Irccs Ca' Granda, Ospedale Maggiore Policlinico, Milan, Italy; 5. Centro Clinico Nemo Ancona, Ancona; 6. IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Clinica Neurologica, Bologna, Italy

P06 - SMN circ4-2b-3 is expressed in nusinersen treated SMA I children and correlates with motor outcomes

MARIKA GUERRA¹, ALBERTO MARINI¹, VITTORIA PAGLIARINI¹, CONSUELO PITOLLI¹, GIORGIA CORATTI^{1,2}, DAVIDE BONVISSUTO¹, CHIARA BRAVETTI^{1,2}, MARIKA PANE^{1,2}, EUGENIO MERCURI^{1,2}, CLAUDIO SETTE¹, MARIA CARMELA PERA^{1,2}

1. Università Cattolica del Sacro Cuore, Rome, Italy; 2. IRCCS Gemelli, Rome, Italy

P07 - Nusinersen impact on cerebrospinal fluid transcriptome of Spinal Muscular Atrophy adult patients

FRANCESCA DRAGONI¹, BARTOLO RIZZO^{1,2}, IRENE MARIA DAINESI^{3,4}, ROSALINDA DI GERLANDO^{1,2}, STEFANO PARRAVICINI^{3,4}, ANGELA LUCIA BERARDINELLI⁴, STELLA GAGLIARDI¹

1. Molecular Biology and Transcriptomic Unit, IRCCS Mondino Foundation, Pavia, Italy; 2. Department of Biology and Biotechnology "L. Spallanzani", University of Pavia, Pavia, Italy; 3. Department of Brain and Behavioral Sciences, University of Pavia, Pavia, Italy; 4. Child and Adolescent Neurology Unit, IRCCS Mondino Foundation, Pavia, Italy

P08 - A case of Spinal Muscular Atrophy with genetic susceptibility for thrombotic microangiopathies

FEDERICA RICCI¹, ANNA SALVALAGGIO¹, ELEONORA GUARNONE¹, ILARIA CAVALLINA¹, FRANCESCA ROSSI¹, ENRICA ROLLE¹, ROSSELLA D'ALESSANDRO¹, TIZIANA MONGINI²

1. Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, University of Turin, Turin, Italy; 2. Department of Neurosciences RLM, Neuromuscular Unit, University of Turin, Italy

P09 - Bladder and bowel dysfunction in spinal muscular atrophy: expanding the multidisciplinary care

FEDERICA RICCI¹, ALESSANDRA SOMÀ¹, ELEONORA GUARNONE¹, ANDREA BUSCAGLIA¹, ROSSELLA D'ALESSANDRO¹, ILARIA CAVALLINA¹, ENRICA ROLLE¹, TIZIANA MONGINI², EUGENIO MERCURI^{3,39}, MARIKA PANE^{3,39}, GIORGIA CORATTI^{3,39}, MARTINA RICCI^{3,39}, ANNA CAPASSO^{3,39}, ADELE D'AMICO⁴, VALERIA SANSONE⁵, CLAUDIO BRUNO⁶, SONIA MESSINA⁷, MICHELA COCCIA⁸, GABRIELE SICILIANO⁹, ELENA PEGORARO¹⁰, MARA TURRI¹¹, MASSIMILIANO FILOSTO^{12,40}, GIACOMO COMI^{13,41}, RICCARDO MASSON¹⁴, LORENZO MAGGI¹⁵, IRENE BRUNO¹⁶, MARIA GRAZIA D'ANGELO¹⁷, ANTONIO TRABACCA¹⁸, VERA VACCHIANO¹⁹, MARIA DONATI²⁰, ISABELLA SIMONE²¹, LUCIA RUGGIERO²², ANTONIO VARONE²³, LORENZO VERRIELLO²⁴, ANGELA BERARDINELLI²⁵, CATERINA AGOSTO²⁶, ANTONELLA PINI²⁷, MARIA ANTONIETTA MAIOLI²⁸, LUIGIA PASSAMANO²⁹, FILIPPO BRIGHINA³⁰, NICOLA CARBONI³¹, MATTEO GARIBALDI³², RICCARDO ZUCCARINO³³, DELIO GAGLIARDI³⁴, SABRINA SILIQUINI³⁵, STEFANO PREVITALI³⁶, DOMENICA TARUSCIO³⁷, STEFANIA BOCCIA³⁸, MARIA CARMELA PERA^{3,39}

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Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Irccs Istituto Giannina Gaslini, Genova, Italy; 7. Department of Clinical and Experimental Medicine, University of Messina; 8. Department of Neurological Sciences, AOU Ospedali Riuniti di Ancona, Ancona, Italy; 9. AOU Pisana, Department of Clinical and Experimental Medicine, Neurology Unit, Pisa, Italy; 10. Neurology Unit, Azienda Ospedale Padova, Padua, Italy; 11. Department of Neurology, Stroke Unit, Bolzano Hospital, Bolzano, Italy; 12. Department of Clinical and Experimental Sciences, University of Brescia, Brescia, Italy; 13. Neurology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; 14. Fondazione IRCCS Istituto Neurologico Carlo Besta, Developmental Neurology Unit, Milan, Italy; 15. Neuroimmunology and Neuromuscular Disorders Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy; 16. Institute for Maternal and Child Health, IRCCS, Burlo Garofolo, Trieste, Italy; 17. Neuromuscular Unit, Scientific Institute IRCCS E. Medea, Bosio Parini, Lecco, Italy; 18. Scientific Institute IRCCS "E. Medea", Unit for Severe Disabilities in Developmental Age and Young Adults (Developmental Neurology and Neurorehabilitation), Brindisi, Italy; 19. UOC Clinica Neurologica, IRCCS Institute of Neurological Sciences of Bologna, Italy; 20. Metabolic Unit, A. Meyer Children's Hospital, Florence, Italy; 21. Neurology Unit, Azienda Ospedaliero-Universitaria, Policlinico Bari "Amaducci", Bari, Italy; 22. Department of Neurosciences, Reproductive Sciences and Odontostomatology, University of Naples Federico II, Naples, Italy; 23. Department of Neurosciences, Pediatric Neurology, Santobono-Pausilipon Children's Hospital, Naples, Italy; 24. Neurology Unit, Department of Neurosciences, University Hospital Santa Maria della Misericordia, Udine, Italy; 25. Department of Child Neuropsychiatry, Fondazione Istituto Neurologico Nazionale C Mondino, IRCCS, Pavia, Italy; 26. Dipartimento di Salute della Donna e del Bambino, Università di Padova, Padua, Italy; 27. Irccs Istituto delle Scienze Neurologiche di Bologna, UOC Neuropsichiatria Infantile, Bologna, Italy; 28. Centro Sclerosi Multipla, P.O. Binaghi, Assl Cagliari, Cagliari, Italy; 29. Cardiomyology and Medical Genetics, AOU and University of Campania Luigi Vanvitelli, Naples, Italy; 30. Section of Neurology, Department of Biomedicine, Neuroscience, and Advanced Diagnostics (BIND), University of Palermo, Palermo, Italy; 31. Neurology Department, Hospital San Francesco of Nuoro, Nuoro, Italy; 32. Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), Sapienza University of Rome, Sant'Andrea Hospital, Rome, Italy; 33. Neuromuscular Omnicentre (Nemo) Trento-Fondazione Serena Onlus, Pergine Valsugana, Italy; 34. Pediatric Neurology Unit, Pediatric Hospital "Giovanni XXIII", Bari, Italy; 35. Child Neuropsychiatry Unit, Paediatric Hospital G Salesi, Ancona, Italy; 36. Institute of Experimental Neurology (INSPE), Division of Neuroscience, IRCCS San Raffaele Scientific Institute, Milan, Italy; 37. National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy; 38. Sezione di Igiene, Istituto di Sanità Pubblica, Università Cattolica del Sacro Cuore, Rome, Italy; 39. Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy; 40. Nemo-Brescia Clinical Center for Neuromuscular Diseases, Brescia, Italy; 41. Dino Ferrari Center, Department of Pathophysiology and Transplantation, University of Milan, Italy

P10 - Map the SMA protocol: a Machine-learning based Algorithm to Predict Therapeutic response in SMA

LAURA ANTONACI, GIORGIA CORATTI, ALBERTO MARINI, CARLOTTA MASCIOCCHI
Fondazione Policlinico Gemelli IRCCS, Rome, Italy

P11 - Serum Creatine Kinase and creatinine levels and clinical features in patient with Spinal Muscular Atrophy under treatment with nusinersen

MARIA IRENE DAINESI¹, ALICE GARDANI², CARLO ALBERTO QUARANTA¹, ARIANNA IOSCA¹, SARA FUSCO¹, LAURA CARRARO¹, STEFANO PARRAVICINI^{1,2}, ANGELA LUCIA BERARDINELLI²
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Metabolic Myopathies - Mitochondrials

Thursday June 6, 2024 10:15 - 14:00

Chairperson: Guido Primiano (Roma)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P12 - Anaphylaxis and desensitization to alglucosidase alfa in late-onset Pompe Disease ten years later after initial enzyme replacement therapy administration: a possible drug-drug interaction of CGRP inhibiting therapies

ALBERTO LERARIO¹, ELENA ABATI¹, DANIELE VELARDO¹, GIACOMO P. COMI², STEFANIA CORTI¹, MONICA SCIACCO¹

1. Neuromuscular and Rare Disease Unit, Fondazione Irccs Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; 2. Neurology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy.

P13 - Recombinant human acid alpha-glucosidase enzyme replacement therapy in non classic infantile-onset Pompe disease (IOPD). A single-case longitudinal study

MARIA CARMELA OLIVA¹, GABRIELE GIANNOTTA¹, DONATELLA DE GIOVANNI², MARTA RUGGIERO¹, IVANA GALLO¹, ISABELLA FANIZZA¹, RITA FISCHETTO², ANTONIO TRABACCA¹

1. Associazione "La Nostra Famiglia", IRCCS "E. Medea", Scientific Hospital for Neurorehabilitation, Unit for Severe Disabilities in developmental age and young adults (Developmental Neurology and Neurorehabilitation), Brindisi, Italy; 2. Department of Metabolic Diseases and Clinical Genetics, Giovanni XXIII Children Hospital, AOUC, Bari, Italy

P14 - Comprehensive central nervous system (CNS) functional evaluation in Italian children affected by Pompe disease: a longitudinal multicentre study

GLORIA GALLO¹, TIZIANA MONGINI², FEDERICA RICCI¹, ANTONIO TRABACCA³, MARIA CARMELA OLIVA³, GIANCARLO PARENTI⁴, SIMONA FECAROTTA⁴, SABRINA SILIQUINI⁵, FEDERICA DEODATO⁶, BENEDETTA GRECO⁶, ALESSIA GRAZIOSI⁶, SERENA GASPERINI⁷, VIOLA CRESCITELLI⁷, GAIA KULLMANN⁷, RITA MARIA ELISA BARONE⁸, LARA CIRNIGLIARO⁸, FRANCESCA MENNI⁹, FRANCESCA FURLAN⁹, ANDREA CELATO⁹, FEDERICO MONTI⁹, ANGELA BERARDINELLI¹⁰, ARIANNA IOSCA¹⁰, ALICE GARDANI¹⁰, ALBERTO BURLINA¹¹, VINCENZA GRAGNANIELLO¹¹, DANIELA GUERALDI¹¹, CHIARA CAZZORLA¹¹, ELENA PROCOPIO¹², MICHELE SACCHINI¹²

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P15 - Pompe Disease: a treatable condition still underdiagnosedCARLOTTA POZZI¹, BENEDETTA FRANCESCA RATTI DI DESIO LEVI², VALENTINA ROVELLI³

1. Department of Pediatrics, Buzzi Children's Hospital, University of Milan, Milan, Italy;

2. Pediatric Unit, Foundation IRCCS Ca' Granda Ospedale Maggiore Policlinico, University of Milan, Milan, Italy; 3. Clinical Department Of Pediatrics, San Paolo Hospital, Asst Santi Paolo e Carlo, University of Milan, Milan, Italy

P16 - Riboflavin-responsive lipid-storage myopathy: a case reportSARA LOPRIENO¹, FRANCESCA TORRI¹, GABRIELE VADI¹, BEATRICE CIURLI¹, FULVIA BALDINOTTI², MARIA ADELAIDE CALIGO², GRETA ALI³, GIOVANNA CENACCHI⁴, GIULIA RICCI¹, GABRIELE SICILIANO¹

1. Department of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy;

2. Section of Molecular Genetics, Department of Laboratory Medicine, AOU Pisana, Pisa, Italy;

3. Unit of Pathological Anatomy, University of Pisa, Pisa, Italy; 4. Department of Biomedical and Neuromotor Sciences (DIBINEM), University of Bologna, Bologna, Italy

P17 - A novel patient presenting late-onset Polyglucosan Body myopathy due to GYG1 mutations: a case report and review of the literatureNICOLA MOLITIerno¹, DANIELE VELARDO², ELENA ABATI^{1,3}, SARA ANTOGNOZZI³, MICHELA RIPOLONE², SIMONA ZANOTTI², LAURA NAPOLI², PATRIZIA CISCATO², MONICA SCIACCO², GIACOMO PIETRO COMI^{1,3}, STEFANIA PAOLA CORTI^{1,2}, DARIO RONCHI^{1,3}

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P18 - Clinical and genetic features of a cohort of patients with myoadenilate deaminase deficiency: a new mutationCLAUDIA ALBERTI¹, SABRINA LUCCHIARI¹, MARTINA RIMOLDI², FRANCESCO FORTUNATO², DANIELE VELARDO¹, LAURA NAPOLI³, MAURIZIO MOGGIO^{1,3}, NEREO BRESOLIN^{1,2}, GIACOMO PIETRO COMI^{1,2}, STEFANIA CORTI^{1,3}, ELENA ABATI^{1,2}

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3. Neuromuscular and Rare Diseases Unit, Fondazione Irccs Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy

P19 - Cardiac comorbidities in McArdle disease: case report and systematic reviewGABRIELE VADI¹, DOMENIKO HOXHAJ¹, LORENZO BIANCHI², LORENZO FONTANELLI¹, FRANCESCA TORRI¹, GIULIA RICCI¹, GABRIELE SICILIANO¹

1. Department of Neuroscience, Neurological Institute, University of Pisa, Pisa, Italy; 2. Department of Internal Medicine, University of Genova, Genova, Italy

P20 - A case of GSDV with unexplained increased transaminases.MATTIA PORCINO^{1,2}, IGNAZIO GIUSEPPE ARENA^{1,2}, MARIA GRAZIA IGEA FALCONE^{1,2}, CRISTIAN USBERGO¹, CARMELO RODOLICO^{1,2}, ANTONIO TOSCANO¹, OLIMPIA MUSUMECI^{1,2}

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2. Unit of Neurology and Neuromuscular Disorders, University of Messina, Messina, Italy

P21 - Spinal cord involvement in Kearns-Sayre Syndrome: double-trouble or disease-related lesions?

GUIDO URBANO¹, LILIANA VERCELLI¹, MARCO VERCELLINO², CHIARA BOSA³, GIULIO GADALETA¹, GIORGIA BRODINI¹, PAOLA CAVALLA², FEDERICA RICCI⁴, TIZIANA MONGINI¹

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P22 - A complex presentation of mitochondrial myopathy

ANNA RUSSO¹, DARIO ZOPPI¹, EMANUELE CASSANO¹, FLORIANA VITALE¹, ROSARIO RUSSO¹, DENISE CASSANDRINI², CHIARA NESTI², ROSA IODICE¹, FIORE MANGANELLI¹, FILIPPO MARIA SANTORELLI², LUCIA RUGGIERO¹

Department of Neurosciences, Reproductive and Odontostomatological Sciences, University of Naples Federico II, Naples, Italy; 2. Molecular Medicine, IRCCS Fondazione Stella Maris, Pisa, Italy

P23 - FGF-21 and GDF-15 AS circulating biomarkers for Mitochondrial Diseases. Validation in one-thousand plasma samples.

SILVIA MARCHETI, ANNA ARDISSONE, KRISZTINA EINVAG, DANIELE SALA, ALESSIA CATANIA, COSTANZA LAMPERTI

IRCCS Neurological Institute Carlo Besta, Milan, Italy

P24 - Prompt diagnosis and treatment of TK2 mitochondrial myopathy: a case report.

DARIA DIODATO¹, MICHELE TOSI¹, MICHELA CATTERUCCIA¹, FRANCESCO NICITA¹, MARGHERITA VERARDO¹, FRANCESCA CUMBO¹, LUCA BOSCO², FABIANA FATTORI², ALESSANDRA TORRACO², ROSALBA CARROZZO², LORENA TRAVAGLINI², ENRICO BERTINI¹, ADELE D'AMICO¹

1. Muscular and Neurodegenerative Disease Unit, Ospedale Pediatrico Bambino Gesù; 2. Genetics Unit, Ospedale Pediatrico Bambino Gesù, Rome, Italy

P25 - Clinical characteristics and outcome of Mitochondrial Parkinsonism in patients with PMM

IGNAZIO GIUSEPPE ARENA, MATTIA PORCINO, GRAZIA MARIA IGEA FALCONE, CRISTIAN USBERGO, ALESSIA PUGLIESE, CARMELO RODOLICO, OLIMPIA MUSUMECI
Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy

Myasthenia Gravis I

Thursday - June 6, 2024 10:15 - 14:00

Chairperson: Liliana Vercelli (Torino)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P26 - Rescue from COVID-19 infection after precipitation of MG crisis in late-onset myasthenia gravis.

CORRADO ANGELINI

University of Padua, Padua, Italy

P27 - Anti-FcRn treatment for generalized Myasthenia Gravis: a real word experience with efgartigimod in AChR-Ab positive patients.

RITA FRANGIAMORE¹, ELENA RINALDI¹, FIAMMETTA VANOLI¹, SILVIA BONANNO¹, FRANCESCA ANDREETTA¹, LORENZO MAGGI¹, ALESSANDRO PINNA², ROBERTO ARNABOLDI², CARLO ANTOZZI¹, RENATO MANTEGAZZA¹

1. UOC Neurology IV - Neuroimmunology And Neuromuscular Diseases Unit, Fondazione IRCCS ISTITUTO Neurologico Carlo Besta, Milan, Italy; 2. Argenx Italy, S.R.L., Milan, Italy

P28 - Subcutaneous efgartigimod PH20 treatment in participants with generalized myasthenia gravis in ADAPT-SC+: interim analyses on quality of life, efficacy, tolerability, and long-term safety

TUAN VU¹, LAURA FIONDA², JAMES F. HOWARD³, DENIS KOROBKO⁴, MAREK SMILOWSKI⁵, FIEN GISTELINCK⁶, SOPHIE STEELAND⁶, JAN NOUKENS⁷, JANA PODHORNÁ⁶, YUEBING LI⁸, KIMIAKI UTSUGISAWA⁹, FRANCESCO SACCÀ¹⁰, HEINZ WIENDL¹¹, JAN L. DE BLEECKER¹², RENATO MANTEGAZZA¹³

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P29 - A rare case of a late-onset congenital myasthenic syndrome associated to co-occurrence of variants in the LRP4 and CHRND genes.

ALESSIA PERNA¹, FILIPPO MARIA SANTORELLI², ELISA COLAIZZO³, SILVIA LA CESA¹, VITTORIO RISO¹, LUDOVICO LISPI¹, ANTONIO PETRUCCI¹

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P30 - Early real-life experience with efgartigimod in AChR-seropositive generalized myasthenia gravis

VINCENZO DI STEFANO, NICASIO RINI, PAOLO ALONGE, SOFIA CAMPO, ALESSIA BONAVENTURA, ANTONINO LUPICA, ANGELO TORRENTE, FILIPPO BRIGHINA
Department of Biomedicine, Neuroscience, and Advanced Diagnostics (BIND), University of Palermo, Palermo, Italy

P31 - Steroid-sparing effect of Eculizumab therapy in patients with anti-acetylcholine receptor antibody-positive generalized myasthenia gravis

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2. Università Cattolica del Sacro Cuore, Rome, Italy

P32 - Long-term efficacy and safety of ravulizumab, a long-acting terminal complement inhibitor, in adults with anti-acetylcholine receptor antibody-positive generalized Myasthenia Gravis: Final results from the Phase 3 CHAMPION MG open-label extension

Presented by: CARLO ANTOZZI²

TUAN VU¹, RENATO MANTEGAZZA², DJILLALI ANNANE³, MASAHISA KATSUNO⁴, ANDREAS MEISEL⁵, MICHAEL NICOLLE⁶, VERA BRIL⁷, RASHA AGUZZI⁸, GLEN FRICK⁸, JAMES HOWARD JR⁹, CHAMPION MG STUDY GROUP

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4. Nagoya University Graduate School of Medicine, Nagoya, Japan; 5. Charité

Universitätsmedizin Berlin, Berlin, Germany; 6. London Health Sciences Centre, London, ON,

Canada; 7. Ellen & Martin Prosserman Centre for Neuromuscular Diseases, University Health

Network, University of Toronto, Toronto, ON, Canada; 8. Alexion, AstraZeneca Rare Disease,

Boston, MA, USA; 9. The University of North Carolina, Chapel Hill, NC, USA

P33 - Clinically meaningful improvement in physical fatigue and muscle weakness fatigability with rozanolixizumab: post hoc analysis of MG symptoms pro responder rate in the MycarinG study

FIAMMETTA VANOLI^{1,4}, JULIAN GROSSKREUTZ², ALI AAMER HABIB³, RENATO MANTEGAZZA⁴, JOHN VISSING⁵, TUAN VU⁶, MARION BOEHNLEIN⁷, BERNHARD GREVE⁷, FIONA GRIMSON⁸, ASHA HAREENDRAN⁸, VERA BRIL⁹

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FL, USA; 7. UCB Pharma, Monheim, Germany; 8. UCB Pharma, Slough, UK; 9. University Health

Network, Toronto, ON, Canada

P34 - AChR-positive generalized Myasthenia Gravis patients unresponsive to new targeted molecular therapies: a single-centre case series

DEMETRIO MARANDO¹, STEFANIA MORINO¹, LUCA LEONARDI¹, LAURA TUFANO², ANTONIO LAULETTA², ELENA ROSSINI¹, FRANCESCA FORCINA¹, MATTEO GARIBALDI^{1,2}, GIOVANNI ANTONINI², LAURA FIONDA¹

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2. Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), Sapienza University of Rome, Rome, Italy

P35 - Differences in clinical severity according to MG ADL score in patients with Myasthenia Gravis.

GIULIA GRECO, LAURA BOFFA, ERICA FREZZA, MARIANGELA GOGLIA, GIULIA NARDINO, ROBERTO MASSA

Neuromuscular Diseases Unit, Policlinico e Università degli Studi di Roma Tor Vergata, Roma, Italy

P36 - Efgartigimod in AChR-positive generalized Myasthenia Gravis: a real-world monocentric longitudinal experience.

BENEDETTA SORRENTI¹, CAMILLA MIRELLA MARIA STRANO¹, CHRISTIAN LAURINI¹, ADELE RATTI¹, LUCA BOSCO², MASSIMO FILIPPI³, YURI MATTEO FALZONE¹, STEFANO CARLO PREVITALI⁴

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Myasthenia Gravis II

Thursday - June 6, 2024 10:15 - 14:00

Chairperson: Michelangelo Maestri Tassoni (Pisa)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P37 - Seasonal Variation in Myasthenia Gravis Incidence

SILVIA FALSO¹, PIETRO ZARA², SOFIA MARINI¹, MARIANGELA PUCI³, ELEONORA SABATELLI⁴, GIOVANNI SOTGIU³, MARTINA MARINI¹, GREGORIO SPAGNI⁵, AMELIA EVOLI¹, PAOLO SOLLA², RAFFAELE IORIO⁴, ELIA SECHI²

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P38 - AChR-Seropositive Myasthenia Gravis in Muscular Dystrophy: diagnostic pitfalls and clinical management challenges

ANNA ROSA AVALLO¹, LILIANA BEVILACQUA¹, PAOLO ALONGE², ANTONINO LUPICA², SIMONA MACCORA², SONIA AMABILE³, FILIPPO BRIGHINA², VINCENZO DI STEFANO², CLAUDIA VINCIGUERRA¹

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P39 - Anti-SRP immune-mediated necrotizing myopathy in a patient with anti-AChR Myasthenia Gravis and thymoma.

VINCENZO CARLOMAGNO, ALESSANDRA CICIA, ELEONORA TORCHIA, SARA BORTOLANI, RAFFAELE IORIO, ENZO RICCI, MASSIMILIANO MIRABELLA, MATTEO LUCCHINI
Fondazione Policlinico Gemelli, Università Cattolica del Sacro Cuore, Roma, Italy

P40 - Frequency of LRP4 antibodies in a consecutive cohort of suspected Myasthenia Gravis patients.

PIETRO BUSI¹, STEFANO MASCIOCCHI¹, SILVIA SCARANZIN², CHIARA MORANDI², ANTONIO MALVASO¹, MATTEO SCUCCHI¹, ELISABETTA ZARDINI², DIEGO FRANCIOTTA², MATTEO GASTALDI²

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P41 - Corticosteroid dose tapering in patients with generalised Myasthenia Gravis on zilucoplan: an interim analysis of RAISE-XT

SOFIA MARINI¹, MIRIAM FREIMER², CHANNA HEWAMADDUMA^{3,9}, MARIA ISABEL DA SILVA LEITE⁴, RAPHAËLLE BEAU LEJDSTROM⁵, BABAK BOROOJERDI⁶, FIONA GRIMSON⁷, NATASA SAVIC⁵, JAMES FRANCIS HOWARD JR⁸

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P42 - Early real-life experience of complement factor C5 inhibitors in refractory generalized Myasthenia Gravis

NICASIO RINI, PAOLO ALONGE, ANTONIO PEZZANO, GIUSEPPE DI MARTINO, UMBERTO QUARTETTI, ANGELO TORRENTE, ANTONINO LUPICA, FILIPPO BRIGHINA, VINCENZO DI STEFANO

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P43 - Kaposi's sarcoma in Myasthenia Gravis patient in oral steroids treatment: two case reports

ADELE BARBACCIA¹, ALESSIA PUGLIESE¹, FIAMMETTA BIASINI¹, ALBA MIGLIORATO², CARMELO RODOLICO¹

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2. Department of Biomedical and Dental Sciences and Morpho-Functional Imaging, University of Messina, Italy

P44 - Long lasting effectiveness of Efgartigimod in anti-acetylcholine receptor antibodies positive Myasthenia Gravis.

GIORGIA CAMERA, PAOLO PAONE, EMANUELA AGAZZI, DARIO ALIMONTI, MANLIO SGARZI
Asst Papa Giovanni XXIII, Bergamo, Italy

P45 - Rapid regression of bulbar symptoms in Myasthenia Gravis refractory clinical relapse after eculizumab infusion as rescue therapy

GIORGIA CAMERA¹, PAOLO PAONE¹, EMANUELA AGAZZI¹, PAOLA BAZZI², DARIO ALIMONTI¹, MANLIO SGARZI¹

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P46 - Myasthenia Gravis as part of the graft-versus-host disease spectrum

LILIANA BEVILACQUA, ANNA ROSA AVALLONE, GIUSEPPE PISCOSQUITO, ANIELLO IOVINO, PAOLO BARONE, CLAUDIA VINCIGUERRA

Neurology Unit, University Hospital "San Giovanni Di Dio e Ruggi D'aragona", Salerno, Italy

P47 - Epilepsy and myasthenia gravis, clues of a complex interplay: case series and review of the literature

FRANCESCA FORCINA, ANDREA CASCIANELLI, ANTONIO LAULETTA, ELENA ROSSINI, LUCA LEONARDI, MATTEO GARIBALDI, STEFANIA MORINO, LAURA TUFANO, MARCO SALVETTI, PAOLO TISEI, GIOVANNI ANTONINI, LAURA FIONDA

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Congenital Myopathies

Thursday - June 6, 2024 10:15 - 14:00

Chairperson: Chiara Fiorillo (Genova)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P48 - Focal myositis in a patient with anti-AChR positive Myasthenia Gravis

ALESSANDRA CICIA, RAFFAELE IORIO, VINCENZO CARLOMAGNO, SILVIA FALSO, SOFIA MARINI, MASSIMILIANO MIRABELLA, MATTEO LUCCHINI

Università Cattolica del Sacro Cuore - Fondazione Policlinico Agostino Gemelli, Roma, Italy

P49 - When "Ulysses struggles to find his way home": a genetic odyssey of a challenging case of congenital myopathy

ANDREA BARP¹, ISABELLA MORONI², MARCELLA NERI³, FERNANDA FORTUNATO³, ILARIA VIVALDI³, RICCARDO ZUCCARINO¹, LORENZO MAGGI⁴, ELIANA IANNIBELLI⁴, MATTEO DE IORIO⁵, MARIA IASCONI⁶, SILVIA MAZZOLA⁷, FRANCESCA GUALANDI³

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P.50 - A novel variant in the HSPB3 gene presenting as congenital myopathy.

A case report

MICHELE TOSI¹, MARGHERITA VERARDO¹, LUCA BOSCO^{2,3}, FABIANA FATTORI³, SERENA CARRA⁴, ALESSANDRO ROSA^{5,7}, MARCO CIRILLO⁶, ADELE D'AMICO¹, ENRICO BERTINI¹

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P51 - A new variant in MYH2 gene in an Italian patient expands the clinical spectrum of Congenital Myopathy-6 with ophthalmoplegia (CMYP6)

SIMONA ZANOTTI¹, DARIO RONCHI^{2,3}, LAURA NAPOLI¹, MICHELA RIPOLONE¹, PATRIZIA CISCATO¹, ROBERTO DEL BO³, FRANCESCA MAGRI³, GIACOMO PIETRO COMI^{2,3}, STEFANIA CORTI^{1,2}, MONICA SCIACCO¹

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P52 - Recurrent respiratory infections in congenital myopathy due to mutation of PYROXD1 gene

AMANDA FERRERO, LUCIA PETTINARI, LUCA COSTANTINI, FEDERICA FELLONI, SARA LUPONE, NOEMI GIRONELLA, MICHELA COCCIA

Neuromuscular Omnicentre (NEMO), AOU delle Marche, Ancona, Italy

P53 - Atypical BAG3-related adult-onset clinical presentation

NICOLA MOLITIERNO¹, DANIELE VELARDO², ELENA ABATI^{1,3}, SARA ANTOGNOZZI³, MICHELA RIPOLONE², SIMONA ZANOTTI², LAURA NAPOLI², PATRIZIA CISCATO², MONICA SCIACCO², STEFANIA PAOLA CORTI^{1,2}, GIACOMO PIETRO COMI^{1,3}, DARIO RONCHI^{1,3}

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P54 - Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (Dista-Myo): longitudinal results

SARA BORTOLANI^{1,2}, ALEX VICINO^{3,4}, MANUELA GAMBELLA⁵, LUISA VILLA⁵, MARCO RABUFFETTI⁶, ALBERTO MARZEGA⁶, VALERIO TROMBETTA¹, MARTA CHELI⁴, AURORA PARROTTA⁵, ENRICA ROLLE², ELEONORA TORCHIA⁷, BEATRICE RAVERA⁷, MAURO MONFORTE¹, JEAN-YVES HOGREL⁸, ENZO RICCI^{1,7}, TIZIANA ENRICA MONGINI², SABRINA SACCONI⁵, LORENZO MAGGI⁴, GIORGIO TASCA⁹

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P55 - SEP1-related myopathy with acute respiratory onset in middle age: a clinical and genetic study

BARBARA RISI¹, FILOMENA CARIA¹, SIMONA DAMIOLI¹, BEATRICE LABELLA^{2,4}, ENRICA BERTELLA¹, GIORGIA GIOVANNELLI¹, LUCIA FERULLO², EMANUELE OLIVERI², MATTIA BUGATTI³, CARLA BARONCHELLI³, LORIS POLI⁴, ALESSANDRO PADOVANI^{2,4}, MASSIMILIANO FILOSTO^{1,2}

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P56 - An integrated approach to monitoring disease activity in inflammatory idiopathic myopathies

LUCA BOSCO¹, ADELE RATTI¹, CHRISTIAN LAURINI¹, BENEDETTA SORRENTI¹, CAMILLA MIRELLA MARIA STRANO¹, VALENTINA CANTI², PATRIZIA ROVERE QUERINI², ANTONIO LAURETTA³, MATTEO GARIBALDI³, ALEX VICINO⁴, LORENZO MAGGI⁴, VINCENZO CARLOMAGNO⁵, MATTEO LUCCHINI⁵, MASSIMILIANO MIRABELLA⁵, MASSIMO FILIPPI¹, UBALDO DEL CARRO⁶, STEFANO CARLO PREVITALI¹

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P57 - RYR1-related myopathies: clinical clues and outcome of a paediatric cohort

ADELE D'AMICO¹, MICHELA CATTERUCCIA¹, DARIA DIODATO¹, IRENE MIZZONI¹, GIACOMO DE LUCA¹, ADELINA CARLES², FABIANA FATTORI³, LUCA BOSCO³, MICHELE TOSI¹, ENRICO BERTINI¹

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3. Laboratory of Medical Genetics, Translational Cytogenomics Research Unit, Bambino Gesù Children Hospital, Rome, Italy

P58 - MYH7-related myopathy: an Italian family with atypical phenotype

DOMENICA ZAINO¹, MELISSA BELLINI², PAOLO IMMOVILLI¹, ROBERTO PARISI³, NICOLA MORELLI⁴, GIACOMO BIASUCCI^{2,5}, CHIARA TERRACCIANO¹

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5. Department of Medicine and Surgery, University of Parma, Parma, Italy

P59 - Progressive myopathy and polyneuropathy

EMANUELA AGAZZI, MANLIO SGARZI, GIORGIA CAMERA, PAOLO PAONE, DARIO ALIMONTI
ASST Papa Giovanni XXIII Hospital, Bergamo, Italy

Duchenne Muscular Dystrophy

Friday - June 7, 2024 10:30 - 13:30

Chairperson: *Francesca Magri (Milano)*

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P60 - Wearable technologies for the biomechanical analysis of motor disfunctions in DMD patients.

ROSSELLA D'ALESSANDRO¹, ELISA PANERO¹, ILARIA CAVALLINA¹, ENRICA ROLLE¹, FRANCESCA ROSSI¹, FRANCESCA RE¹, FRANCESCA SERTORI¹, LAURA GASTALDI², TIZIANA MONGINI³, FEDERICA RICCI¹

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P61 - DMD deletions underlining asymptomatic and mild dystrophinopathies: literature review highlights phenotype-related mutation clusters and provides insights about genetic mechanisms, etiopathogenesis and prognosis

FERNANDA FORTUNATO, LAURA TONELLI, MARIANNA FARNE', RITA SELVATICI, ALESSANDRA FERLINI

Unit of Medical Genetics, Department of Medical Sciences, University of Ferrara, Ferrara, Italy

P62 - Transcriptomic profile of skeletal muscle biopsies from Duchenne and Becker Muscular Distrophy patients

BARTOLO RIZZO^{1,2}, FRANCESCA DRAGONI², MARIA IRENE DAINESI^{3,5}, ROSALINDA DI GERLANDO^{1,5}, STEFANO PARRAVICINI^{3,5}, ANGELA LUCIA BERARDINELLI⁴, STELLA GAGLIARDI⁵

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P63 - Ankle range of motion asymmetry and motor functional tests: a retrospective analysis of 24 months in ambulant boys with Duchenne Muscular Dystrophy (DMD)

ALICE GARDANI¹, VALERIA VACCHINI¹, MARIA IRENE DAINESI², ARIANNA IOSCA², SARA FUSCO², STEFANO PARRAVICINI¹, ANGELA LUCIA BERARDINELLI¹

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P64 - Motor assessment in DMD: introducing a Novel Parameter usable after LoA

ARIANNA IOSCA¹, ALICE GARDANI², SARA FUSCO¹, MARIA IRENE DAINESI¹, LAURA CARRARO¹, CARLO ALBERTO QUARANTA¹, STEFANO PARRAVICINI^{1,2}, ANGELA LUCIA BERARDINELLI²

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P65 - Clinical features in a large cohort of adults with Duchenne Muscular Dystrophy treated with glucocorticoids

MARIANELA SCHIAVA¹, ROBERT MUNI LOFRA¹, JOHN BOURKE¹, JORDI DÍAZ-MANERA¹, MEREDITH JAMES¹, MAHA ELSEED¹, MONIKA MALINOVA¹, JASSI MICHEL-SODHI¹, DIONNE MOAT¹, ELISABETTA GHIMENTON¹, MICHELLE MCCALLUM¹, CARLA FLORENCIA BOLAÑO DÍAZ¹, ANNA MAYHEW¹, KAREN WONG¹, MARK RICHARDSON¹, GIORGIO TASCA¹, GAIL EGLON¹, MICHELLE EAGLE², CATHY TURNER¹, EMMA HESLOP¹,

VOLKER STRAUB¹, CHIARA MARINI BETTOLO¹, MICHELA GUGLIERI¹

1. John Walton Muscular Dystrophy Research Centre, Translational And Clinical Research Institute, Newcastle University, Newcastle Upon Tyne, UK; 2. Atom International Limited, Gateshead, UK.

P66 - MITRACLIP implantation in a patient with Duchenne Muscular Dystrophy

CONCETTA PALERMO¹, DANIELA LEONE¹, BIANCA BUCHIGNANI¹, PRISCILLA LAMENDOLA², ANTONELLA LOMBARDO^{2,3}, GAETANO LANZA^{2,3}, FRANCESCO BURZOTTA^{2,3}, EUGENIO MERCURI^{1,3}, CARLO TRANI^{2,3}, MARIKA PANE^{1,3}

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P67 - Sleep Disordered Breathing in a cohort of patients with Duchenne Muscular Dystrophy: a retrospective analysis and future perspectives

SARA FUSCO¹, STEFANO PARRAVICINI², ARIANNA IOSCA¹, MARIA IRENE DAINESI¹, LAURA CARRARO¹, CARLO ALBERTO QUARANTA¹, MICHELE TERZAGHI³, ANGELA BERARDINELLI²

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P68 - The DMD Hub Central Recruitment Database, a model for effective recruitment to Rare Disease clinical trials

Presented by: PIETRO RIGUZZI²

MICHELA GUGLIERI¹, PHILLIP CAMMISH¹, PIETRO RIGUZZI^{1,2}, ALEX JOHNSON³, KERRY SHIPPEY³, KATE ADCOCK⁴, SUZANNE GLOVER⁵, EMILY REUBEN³, MARIA ELENA FARRUGIA⁶, ROSALINE QUINLIVAN⁷, TRACEY WILLIS⁸, ANNE-MARIE CHILDS⁹, MARIACRISTINA SCOTO¹⁰, CATHY TURNER¹, MEGAN MCNIFF¹, DAN RISEBOROUGH¹, VOLKER STRAUB¹, EMMA HESLOP¹

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P69 - Impact of vamorolone, prednisone and placebo on linear growth in the VISION-DMD (VBP15-004) study, as measured by changes in height over 6 months

Presented by: LUCA BELLO

ANA DE VERA¹, RAOUL ROOMAN¹, ERIC P HOFFMAN², PAULA R CLEMENS³, MICHELA GUGLIERI⁴, VBP15-004 INVESTIGATORS

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P70 - Evaluation of behavioural problems in the VISION-DMD study of vamorolone vs prednisone in Duchenne muscular dystrophy

Presented by: LUCA BELLO

ERIK HENRICSON¹, ANA DE VERA², MIKA LEINONEN²

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P73 - Givinostat in DMD: results of the Epidys study with particular attention to NSAA

EUGENIO M. MERCURI¹, CLAUDIA BROGNA², MAH JEAN³, NATHALIE GOEMANS⁴, ERIC NIKS⁵, SARA CAZZANIGA S⁶, NICOLETTA COCEANI⁶, PAOLO BETTICA P⁶, CRAIG MCDONALD⁷

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P74 - Givinostat in DMD: results of the Epidys Study with particular attention to MR measures of muscle fat fraction

KRISTA VANDENBORNE¹, REBECCA WILLCOCKS¹, GLENN WALTER², SEAN FORBES³, SARA CAZZANIGA⁴, PAOLO BETTICA⁴, EUGENIO M. MERCURI⁵, CRAIG MCDONALD⁶
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P75 - Preclinical assessment of the effects of the growth hormone secretagogue JMV2894 in the D2-mdx mouse model of Duchenne Muscular Dystrophy

MANUEL MARINELLI¹, PAOLA MANTUANO¹, ORNELLA CAPELLARI¹, BRIGIDA BOCCANEGR¹, LISAMAURA TULIMIERO¹, ANTONIETTA MELE¹, DANIELA TRISCIUZZI¹, ENRICA CRISTIANO¹, ELENA CONTE¹, ELENA BRESCIANI², ANTONIO TORSELLO², SEVERINE DENOYELLE³, ORAZIO NICOLOTTI¹, ANTONELLA LIANTONIO¹, ANNAMARIA DE LUCA¹

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Myotonic Dystrophies

Friday - June 7, 2024 10:30 - 13:30

Chairperson: *Antonio Trabacca (Brindisi)*

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P76 - Initial Data from the ACHIEVE Trial of DYNE-101 in adults with Myotonic Dystrophy type 1 (DM1)

MARIKA PANE¹, GUILLAUME BASSEZ², JORDI DIAZ-MANERA³, JAMES B. LILLEKER⁴, BAZIEL VAN ENGELLEN⁵, RICHARD H ROXBURGH⁶, BENEDIKT SCHOSER⁷, CHRISTOPHER TURNER⁸, CHRIS MIX⁹, SOMA RAY⁹, BAOGUANG HAN⁹, WILDON FARWELL⁹, DANIEL WOLF⁹, ERWAN DELAGE⁹, VALERIA SANSONE¹⁰

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P77 - Low ischemic stroke prevalence despite high burden of cardiovascular risk factors in Myotonic Dystrophy type 1 (DM1): data from a retrospective, observational cohort study

SALVATORE ROSSI^{1,2}, ANTONIO FUNCIS², GIANMARCO DALLA ZANNA², AURELIA ZAULI³, CRISTINA SANCRICCA^{1,3}, PIETRO CALIANDRO^{2,3}, GABRIELLA SILVESTRI^{2,3}

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P78 - Gait parameters / cognitive function correlation in a cohort of patients with Myotonic Dystrophy type 1: a single-center sensor-based gait analysis

BARBARA RISI¹, ANDREA PILOTTO^{2,3,4}, ANDREA RIZZARDI^{2,4}, ELISABETTA FERRARI¹, BEATRICE LABELLA^{2,4}, CINZIA ZATTI^{2,4}, CLINT HANSEN⁵, ROBBIN ROMIJNDERS⁵, FILOMENA CARIA¹, SIMONA DAMIOLI¹, ENRICA BERTELLA¹, LORIS POLI⁴, LUCIA FERULLO^{2,4}, EMANUELE OLIVIERI^{2,4}, WALTER MAETZLER⁵, ALESSANDRO PADOVANI^{2,4}, MASSIMILIANO FILOSTO^{1,2}

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P79 - Functional characterization of the Nav1.4 sodium channel mutation, p. L689F, found in a young boy suffering from myotonia permanens.

CONCETTA ALTAMURA¹, CARMEN CAMPANALE¹, PAOLA LAGHETTI¹, ILARIA SALTARELLA¹, DAMIEN STERNBERG², SAVINE VICART², JEAN-FRANCOIS DESAPHY¹

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P80 - Irisin and brain-derived neurotrophic factor in Myotonic Dystrophy type 1

ERICA FREZZA¹, MARIANGELA GOGLIA¹, GIULIA GRECO¹, VIETRI GIOVANNI¹, ILARIA PETITTA¹, FRANCESCO GRUOSSO¹, GIULIA NARDINO¹, LAURA BOFFA¹, MARZIA NUCCETELLI², ROBERTO MASSA¹

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P81 - Assessing the phenotype and genotype of a wide Italian DM1 and DM2 cohort: results from the Italian Myotonic Dystrophies National Registry

VIRGINIA IACOBELLI¹, MARTINA RIMOLDI², SABRINA LUCCHIARI¹, SERENA PAGLIARANI², GRAZIA D'ANGELO³, ROCCO LIGUORI^{4,14}, FRANCESCO OTTAVIO LOGULLO⁵, ELENA PEGORARO⁶, VALERIA SANSONE⁷, LUCIO SANTORO⁸, MARINA SCARLATO⁹, GABRIELE SICILIANO¹⁰, GABRIELLA SILVESTRI¹¹, GIUSEPPE VITA¹², GIACOMO P. COMI^{2,13}, STEFANIA CORTI^{13,17}, GIOVANNI MEOLA^{15,18}, ELENA ABATI^{2,13}

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P82 - Assessment of sleep quality and sleep disturbances severity perception in a cohort of Myotonic Dystrophy type 1 patients.

MICHELE LOMBARDI, FABIOLA DE MARCHI, MARIA FRANCESCA SARNELLI, CRISTOFORO COMI, LETIZIA MAZZINI

Neurology Department, Ospedale Maggiore della Carità, Novara

P83 - Dropped head in a patient with Myotonic Dystrophy type 1 and Parkinson's disease: a case report and a case-based review.

MICHELE GIOVANNI CROCE¹, FRANCESCA VALENTINO², EMANUELE MICAGLIO³, SARA BENEDETTI³, MATTEO PAOLETTI², GIUSEPPE COSENTINO², SABRINA RAVAGLIA²

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P84 - Sleepy patients with Myothonic Dystrophy tipe 1 do not have a distinct neuropsychological and neuroimaging pattern compared to non sleepy patients

GIOVANNI COLACICCO¹, CAROLA RITA FERRARI AGGRADI¹, MANACORDA FILIPPO², RICCARDI SEBASTIANO², CASIRAGHI JACOPO¹, BARP ANDREA³, STANO SALVATORE³,

LIZIO ANDREA¹, VITALI PAOLO⁴, ZANARDO MORENO⁴, SARDANELLI FRANCESCO⁴,
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P85 - Psoriasis and Myotonic Dystrophy type 1: another cutaneous manifestation of a multisystemic disorder

LAURA TUFANO¹, ELISABETTA BUCCI², GIOVANNI ANTONINI², MATTEO GARIBALDI¹

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Muscular Dystrophies

Friday - June 7, 2024 10:30 - 13:30

Chairperson: Andrea Barp (Trento)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P86 - SNUPN gene defect causing early onset progressive LGMD with myofibrillar features

MONICA TRAVERSO¹, MICHELE IACOMINO², SERENA BARATTO³, ELENA FAEDO⁴, ALESSANDRO GEROLDI⁴, PAOLO UVA⁵, FEDERICO ZARA^{2,4}, PAOLA MANDICH^{4,6}, MARINA GRANDIS^{4,6}, CHIARA FIORILLO^{4,8}

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P87 - Observational study: the quality of life in patients with beta-sarcoglycan, alpha-sarcoglycan, and aamma-sarcoglycan gene mutations

BEATRICE VOLA¹, MASSIMILIANO CERLETTI², CARLES SANCHEZ RIERA³, OREST SEMERYAK⁴, CHAHNEZ CHARFI TRIKI⁵, YVAN TORRENTE⁶

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P88 - IPSCs generation, characterization and differentiation for LGMDR4 patients

YVAN TORRENTE¹, GIULIO POMPILIO², MASSIMILIANO CERLETTI³, CARLES SANCHEZ RIERA⁴, FRANÇOIS GROS-LOUIS⁵, BEATRICE VOLA⁶

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P89 - Muscle and cardiac phenotypes in pediatric titinopathy: characterization of a single centre cohort.

MICHELA CATTERUCCIA¹, DARIA DIODATO¹, FABIANA FATTORI², MARCO SAVARESE³, LUCA BOSCO^{1,2}, ENRICO BERTINI¹, ADELE D'AMICO¹

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P90 - The CFTR corrector C17 as potential treatment for sarcoglycanopathies: pharmacology and efficacy in mouse

ALBERTO BENETOLLO, MARTINA SCANO, SOFIA PARRASIA, LUCIA BIASUTTO, LEONARDO NOGARA, BERT BLAAUW, DORIANNA SANDONA'
University of Padua

P91 - Sarcoglycanopathy with Central Nervous System Involvement: double trouble in pediatric neurology

MARTINA PENZO¹, GIORGIA SEGRE¹, SARA GIBERTINI², LORENZO MAGGI², GIOVANNA ZORZI¹, ISABELLA MORONI¹

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P92 - Clinical characterization of a large monocentre cohort of patients with Becker Muscular Dystrophy

PIETRO RIGUZZI^{1,2}, HOLLY BORLAND¹, JOHN BOURKE¹, CHIARA MARINI BETTOLO¹, MEREDITH JAMES¹, ROBERT MUNI LOFRA¹, JORDI DIAZ-MANERA¹, GIORGIO TASCA¹, MAHA ELSEED¹, MARIANELA SCHIAVA¹, CARLA BOLANO DIAZ¹, JASSI MICHEL-SODHI¹, DIONNE MOAT¹, KAREN WONG¹, ELENA PEGORARO², LUCA BELLO², VOLKER STRAUB¹, MICHELA GUGLIERI¹

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P93 - A novel de novo SYNE2 mutation causing a childhood form of type 5 Emery-Dreifuss Muscular Dystrophy

ESTHER PICILLO¹, ORLANDO PACIELLO², MARIA ELENA ONORE¹, ALBERTO PALLADINO¹, ELIA CESARONE¹, LUIGIA PASSAMANO¹, VINCENZO NIGRO¹, LUISA POLITANO³

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P94 - Longitudinal echocardiographic measures in Becker muscular dystrophy

GIULIANA CAPECE¹, ANGELA PETROSINO¹, ELENA SOGUS¹, PIETRO RIGUZZI¹, CHIARA CALORE², ELENA PEGORARO¹, LUCA BELLO¹

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P95 - Subjective assessment of sleep quality in adult patients with dystrophinopathy: a single centre experience.

EMANUELE OLIVIERI¹, LUCIA FERULLO¹, LEANDRO PURIN¹, BEATRICE LABELLA¹, BARBARA RISI², FILOMENA CARIA², SIMONA DAMIOLI², LORIS POLI³, MARIA PIA PASOLINI³, ALESSANDRO PADOVANI¹, MASSIMILIANO FILOSTO¹

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P96 - Axial involvement in Facioscapulohumeral dystrophy (FSHD): imaging and clinical features.

BEATRICE RAVERA¹, SARA BORTOLANI², ELENA TORCHIA¹, MAURO MONFORTE²,
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Basic Myology - Genetics

Friday - June 7, 2024 10:30 - 13:30

Chairperson: Maria Grazia D'Angelo (Bosisio Parini)

The Presenting Author has 5 min. for the presentation and 2 min. for the discussion

P97 - SGLT2 inhibitors improve skeletal muscle impairment in an animal model of heart failure by modulating glucose and ion homeostasis

ELENA CONTE¹, PAOLA IMBRICI¹, GIORGIA DI NOI¹, KONRAD URBANEK², DONATO CAPPETTA³, LIBERATO BERRINO⁴, ANNAMARIA DE LUCA¹, ANTONELLA DE ANGELIS⁴, ANTONELLA LIANTONIO¹

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P98 - Characterization of cellular differentiation in an in vitro model of neuromuscular junction using a co-culture system: a preliminary study of a drug testing platform

GAIA CARBONE, VITTORIA CANFORA, SABATA PIERNO, GIULIA MARIA CAMERINO
University of Bari, Bari, Italy

P99 - Unraveling a long-standing case through genetic panels: a rare form of congenital slow-progressive neuromuscular condition.

GIULIO GADALETA¹, LILIANA VERCELLI¹, GUIDO URBANO¹, ENRICA ROLLE¹, LOREDANA CHIADO-PIAT¹, FEDERICA RICCI², TIZIANA MONGINI¹

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P100 - Differential exon usage predicts disease course in the wide titinopathy spectrum

MARIA FRANCESCA DI FEO^{1,28}, ALI OGHABIAN¹, ELLA NIPPALA¹, FRANCESCA FORZANO², EDOARDO Malfatti³, CLAUDIA CASTIGLIONI⁴, MATHIAS GAUTEL⁵, HEINZ JUNGBLUTH⁵, ILONA KREY⁶, DAVID GOMEZ ANDRES⁷, ANGELA BRADY⁸, MARIA IASCONI⁹, ANNA CEREDA¹⁰, LIDIA PEZZANI¹⁰, DANIEL NATERA DE BENITO¹¹, ANDRES NASCIMENTO OSORIO¹², BERTA ESTÉVEZ ARIAS¹³, SERGEI KURBATOV¹⁴, TANIA ATTIE-BITACH¹⁵, SHEELA NAMPOOTHIRI¹⁶, ERIN RYAN¹⁷, MICHELLE MORROW¹⁸, SVETLANA GOROKHOVA¹⁹, BRIGITTE CHABROL²⁰, JUHA SINISALO²¹, HELI TOLPPANEN²¹, JOHANNA TOLVA²², FRANCINA MUNELL²³, JESSICA CAMACHO SORIANO²⁴, MARIA ANGELES SANCHEZ DURAN²⁵, MRIDUL JOHARI²⁶, HOMA TAJSHARGHI²⁷, PETER HACKMAN¹, BJARNE UDD¹, MARCO SAVARESE¹

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28. University of Genova, Genova, Italy

P101 - Biallelic HMGCS1 variants in rigid spine syndrome

CHIARA FIORILLO¹, MONICA TRAVERSO¹, MARCELLO SCALA¹, SERENA BARATTO¹, CLAUDIO BRUNO¹, MARINA PEDEMONTE¹, FEDERICA TRUCCO¹, FEDERICO ZARA¹, GINA RAVENSCROFT²

1. IRCCS Istituto Gaslini, Genova, Italy; 2. Harry Perkins Institute of Medical Research, University of Western Australia, Australia

P102 - The 5-year EU project SCREEN4CARE on genetic newborn screening Selection of treatable diseases

MARIANNA FARNÈ¹, FERNANDA FORTUNATO¹, SILVIA MONTANARI¹, ALICE MARGUTTI¹, VITTORIA NAGLIATI¹, RITA SELVATICI¹, CHRISTINA SAIER², JAN KIRSCHNER², STEFAAN SANSEN³, SILVIA OTTOMBRINO⁴, EMANUELE AGOLINI⁴, ANTONIO NOVELLI⁴, ENRICO SILVIO BERTINI⁵, ALDONA ZYGMUNT⁶, ALESSANDRA FERLINI¹

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P103 - Long-read DNA and RNA sequencing improves analyzing large and complex genes: the example of TTN and NEB

RAFAELA OWUSU¹, ELLA NIPPALA¹, ALI OGHABIAN¹, MARIA FRANCESCA DI FEO², MEHARIJI ARUMILLI¹, DAVID GOMEZ ANDRES³, LYDIA SAGAT⁴, VILMA LOTTA LEHTOKARI¹, JESSICA CAMACHO SORIANO³, MARIA ANGELES SANCHEZ DURAN³, FRANCINA MUNELL³, KATARINA PELIN¹, PETER HACKMAN¹, BJARNE UDD¹, MARCO SAVARESE¹

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P104 - Severe epileptic encephalopathy with progressive cerebral and cerebellar atrophy and peripheral neuropathy: a new GEMIN5 phenotype?

DARIA DIODATO¹, GINEVRA ZANNI¹, ALESSANDRA TERRACCIANO¹, PAOLA DE LISO², CONCETTA LUISI², MARCELLO NICETA³, MARCO TARTAGLIA³, ENRICO BERTINI¹, ADELE D'AMICO¹

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P105 - Muscle phosphorylase b kinase deficiency caused by new mutations in the PHKA1 gene.

MARTA CHELI¹, DENISE CASSANDRINI², DANIELE GALATOLO², GAETANO VATTEMI¹, FILIPPO MARIA SANTORELLI², PAOLA TONIN¹

1. Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona, 37134 Verona, Italy; 2. IRCCS Fondazione Stella Maris, Pisa, Italy

P106 - Novel TMEM43 mutations lead to extreme variability of clinical phenotype

GIULIA MARCHETTO¹, DENISE CASSANDRINI², FRANCESCO DALLA BARBA³, MARTA CHELI¹, DIEGO LOPERGOLO⁴, ALESSANDRO MALANDRINI⁴, CARMELO RODOLICO⁵, FILIPPO MARIA SANTORELLI², PAOLA TONIN¹, MARCELLO CAROTTI³, DORIANNA SANDONÀ³, GAETANO VATTEMI¹

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P107 - Neurofilament light chain as clinical plasma biomarker of Charcot-Marie-Tooth disease

CLAUDIA ALBERTI¹, DOMENICA SACCOMANNO¹, ALESSIA ANASTASIA², GRAZIA D'ANGELO¹, MARIA TERESA BASSI¹, LORENZO QUETTI², LUCA SALI², LORENZO BRAMBILLA², ALBERTO ROMANO², GIACOMO PIETRO COMI^{1,2}, FEDERICA RIZZO², STEFANIA CORTI^{1,3}, ELENA ABATI^{1,2}
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P108 - Subacute ataxic sensitive syndrome

EMANUELA AGAZZI, MANLIO SGARZI, GIORGIA CAMERA, PAOLO PAONE, DARIO ALIMONTI
ASST Papa Giovanni XXIII Hospital, Bergamo, Italy

P109 - Optimization of a screening assay for antibody-mediated complement activation (ACA) in autoimmune neurological disorders

PIETRO BUSINARO¹, STEFANO MASCIOCCHI¹, SILVIA SCARANZIN², CHIARA MORANDI², ELISABETTA ZARDINI¹, DIEGO FRANCIOTTA², MATTEO GASTALDI¹
1. Department of Brain and Behavioral Sciences, University of Pavia, Pavia, Italy; 2. Neuroimmunology Research Unit, IRCCS Mondino Foundation, Pavia, Italy.

P110 - Flexibility, Resistance, Aerobic, Movement Execution (FRAME) training program to improve gait capacity in adults with Hereditary Spastic Paraparesis: protocol for a single-cohort feasibility trial.

LEONARDO BOCCUNI, MARCO BORTOLINI, CRISTINA STEFAN, ANDREA MARTINUZZI
Scientific Institute, IRCCS E. Medea, Department Of Conegliano, Treviso, Italy

P111 - Validation of neutrophil assay for detection of dysferlin in a Latin American cohort

RITA BARRESI¹, ANA TOPF², ALEJANDRO GONZALEZ-CHAMORRO², JORDI DIAZ-MANERA², SARAH SHIRA EMMONS³, LAURA RUFIBACH³
1. San Camillo IRCCS, Venezia, Italy; 2. John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, UK; 3. JAIN Foundation, Seattle, WA, USA

P112 - A diagnosis of Neuroacanthocytosis in a patient with a long history of isolated but considerable hyperckemia

ANNA RUSSO, DARIO ZOPPI, MONICA TRAVERSO, CHIARA FIORILLO¹, ROBERTA PIERA BENCIVENGA, CHIARA GRASSO, MARIANGELA IAPOCE, LUCIA RUGGIERO
Department of Neurosciences, Reproductive and Odontostomatological Sciences, University of Naples Federico II, Naples, Italy; 2. Pediatric Neurology and Muscle Disease Unit, IRCCS Istituto Giannina Gaslini, Genova, Italy

P113 - A familial case of ptosis and dysphagia with facial dysmofisms due to a novel KMT2D gene mutation.

FRANCESCA CORTESE¹, MARIANNA BRIENZA¹, LAURA DE GIGLIO¹, ROSALBA CARROZZO², ALESSANDRA TORRACO², FABIANA FATTORI³, LUCA BOSCO⁴, MARIA CONCETTA ALTAVISTA⁵, ELENA MARIA PENNISI¹

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P114 - A novel pathogenic variant in the DYNC1H1 gene: the combining neuromuscular and brain involvement in the disease spectrum of "Dyneinopathies". A case report.

ANGELA LA TEMPA^{1,2,7}, FEDERICA TRENTIN^{1,2,7}, GIULIA GUARDI^{1,2,7}, MELANIA GIANNOTTA², FLAVIA PALOMBO³, AHMED SHEIK MAYE HODMAN⁴, LAURA LICCHETTA⁵, MARIA LUCIA VALENTINO⁵, ROSARIA PLASMATI⁶, DUCCIO MARIA CORDELLI^{2,7}, ANTONELLA PINI²

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P115 - A case of dropped head syndrome and exercise intolerance in a 5 year old boy due to a pathogenic variant in the CACNA1S gene

FEDERICA TRENTIN^{1,2}, ANGELA LA TEMPA^{1,2}, MELANIA GIANNOTTA¹, ROSARIA PLASMATI⁴, FLAVIA PALOMBO⁵, DUCCIO MARIA CORDELLI^{2,3}, ANTONELLA PINI¹

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P116 - New association between the ZASP/LDB3 Pro26Ser variant and inclusion body myositis.

DANIELA PIGA¹, SIMONA ZANOTTI², MICHELA RIPOLONE², LAURA NAPOLI², PATRIZIA CISCATO², SARA GIBERTINI³, LORENZO MAGGI³, FRANCESCO FORTUNATO⁴, ANDREA RIGAMONTI⁵, DARIO RONCHI^{1,4}, MONICA SCIACCO^{1,2}, GIACOMO PIETRO COMI^{1,4}, STEFANIA CORTI^{2,4}

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P117 - HyperCKemia as a potential biochemical feature of Hypokalemic Periodic Paralysis

JACOPO VENANZI¹, NICCOLÒ CAMPAGNA¹, TICCI CHIARA², FERRI LORENZO³, MORRONE AMELIA³, SACCHINI MICHELE², PROCOPIO ELENA²

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P118 - Countermeasures for sarcopenia: effects of BCAA-based formulations on muscle function, histomorphology, and calcium homeostasis in aged mice.

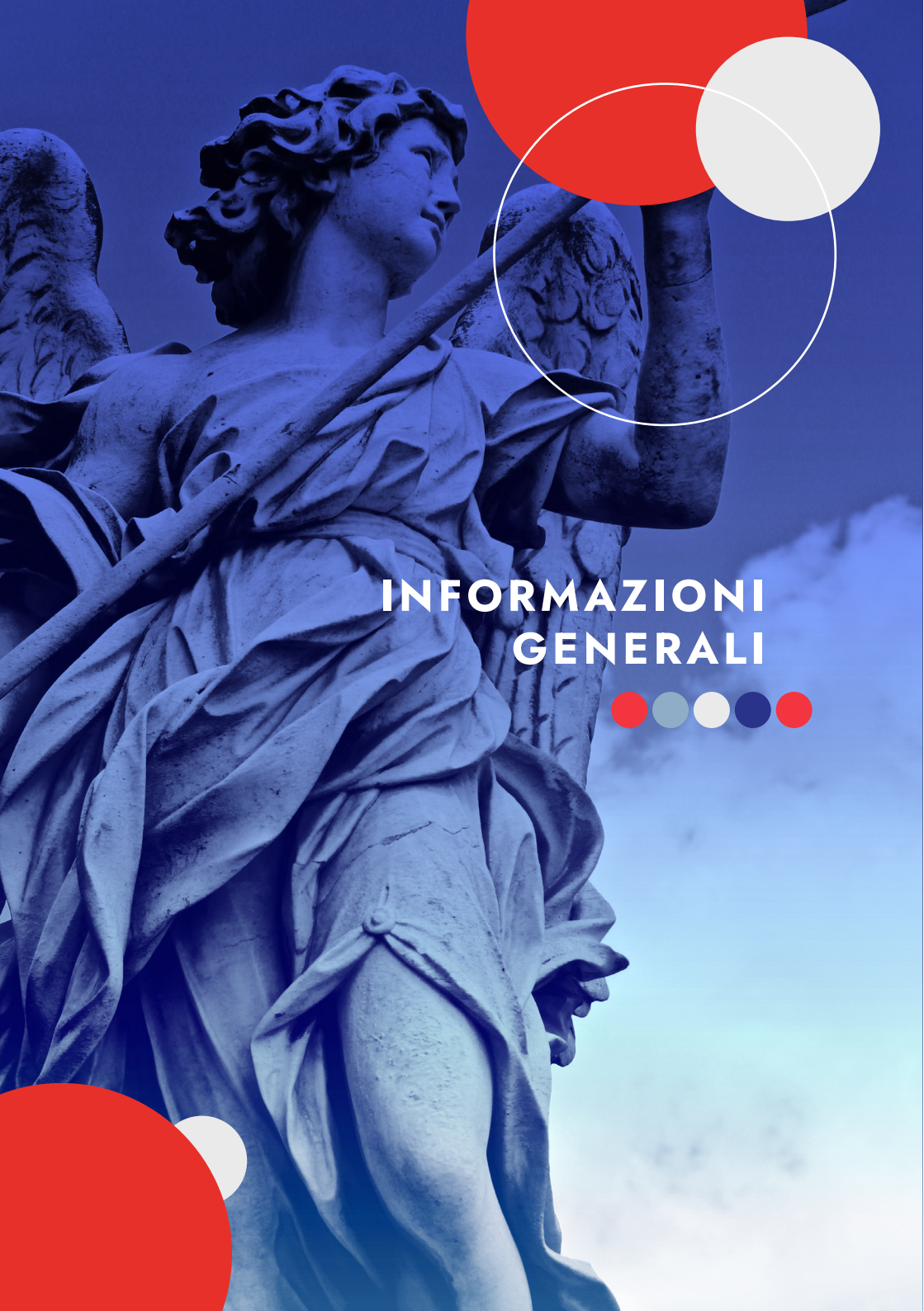
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MICHELA DE BELLIS¹, ANTONIETTA MELE¹, ANTONELLA LIANTONIO¹,
MARCELLO ALLEGRETTI², ANDREA ARAMINI², ANNAMARIA DE LUCA³

1. Università di Bari, Bari, Italy; 2. Dompé Farmaceutici S.p.A, Milan, Italy; 3. Università di Bari, Bari, Italy

P119- Wearable sensors to evaluate and monitor neuromuscular patients in real world environment

ELEONORA DIELLA¹, FABIO STORM¹, LUCA MOLTENI¹, MORENA DELLE FAVE¹, GIULIA
CANELLA¹, GIOVANNI MEOLA², EMILIA BIFFI¹, MARIA GRAZIA D'ANGELO¹

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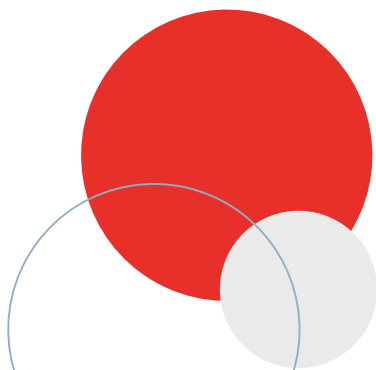
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