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:

Sin
SOCIETÀ ITALIANA DI NEUROLOGIA

Associazione Italiana
Sistema Nervoso Periferico

aisla ONLUS
ASSOCIAZIONE ITALIANA
SCLEROSI LATERALE AMIOTROFICA

AINAT
ASSOCIAZIONE ITALIANA
NEUROLOGI AMBULATORIALI TERRITORIALI

ROMA
Assessorato alle Politiche Sociali e alla Salute

FNOMCeO
Federazione Nazionale degli Ordini
dei Medici Chirurghi e degli Odontoiatri

OMAR
OSSERVATORIOMALATTIERARE

OSSERVATORIO
TERAPIE
AVANZATE

Sumai
ASSOPROF
Sindacato Unico Medicina
Ambulatoriale Italiana
e Professionalità dell'Area Sanitaria

ALTR DOMANI APS-ETS

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CONSULTA
MALATTIE
NEUROMUSCOLARI

FamiglieSMA
Genitori per la Ricerca sull'Atrofia Muscolare Spinale

GRUPPO FAMILIARI
GFB ONLUS
BETA-SARCOGLICANOPATIE

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Insieme per lo studio e la cura
delle malattie mitocondriali ODV

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ASSOCIAZIONE ITALIANA MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

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CONSIGLIO DIRETTIVO

ASSOCIAZIONE ITALIANA MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY

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

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FACULTY

FACULTY

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Costanza Lamperti, Milano
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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 Gualtiero 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

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

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 eliminatoria		
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 muscolare dystrophy research		
20.30	SPEAKER DINNER		

PROGRAM AT A GLANCE

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	POSTER SESSION PART II	
11.00 - 12.30	PLENARY WORKSHOP 4 Mitochondrial diseases: from the emergency department to the new therapies. Avenues and pitfalls		
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		
15.30 - 16.00	COFFEE BREAK		
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		


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 eliminatoria
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

NO CME

15:00 – 16:00 | **SYMPOSIUM – SPONSORED BY ROCHE**
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

GIOVE HALL

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 a15-year follow up study
ESTER LATINI, ALBA CEPELE, MELANIA GUIDA, ILARIA CECCARELLI, CARMELINA CRISTINA ZIRAFA, 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)

GIUNONE HALL 1

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)

GIUNONE HALL 2

16:30 – 17:30 | ORAL COMMUNICATION | METABOLIC MYOPATHIES
Chairpersons: *Massimilano 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)

GIOVE HALL

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)

GIUNONE HALL 1

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 CHELI, 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 CAVALLO, 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

GIOVE HALL

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**

Saturday, 8 June 2024

GIOVE HALL

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)

GIUNONE HALL 1

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)

GIUNONE HALL 2

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 DL 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
Munichen, Tubingen(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); Tubingen (Germany)

GIOVE HALL

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 myopthies
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)

GIUNONE HALL 1

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, Sacrament, Rochester, Gainesville, Norfolk, Washington, Cambridge (USA); London, Welwyn Garden City (UK); Roma (Italia); Tokyo (Japan); Essen (Germany); Basel (Switezerland)

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 GORGOGLIONE, 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, Fearra (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

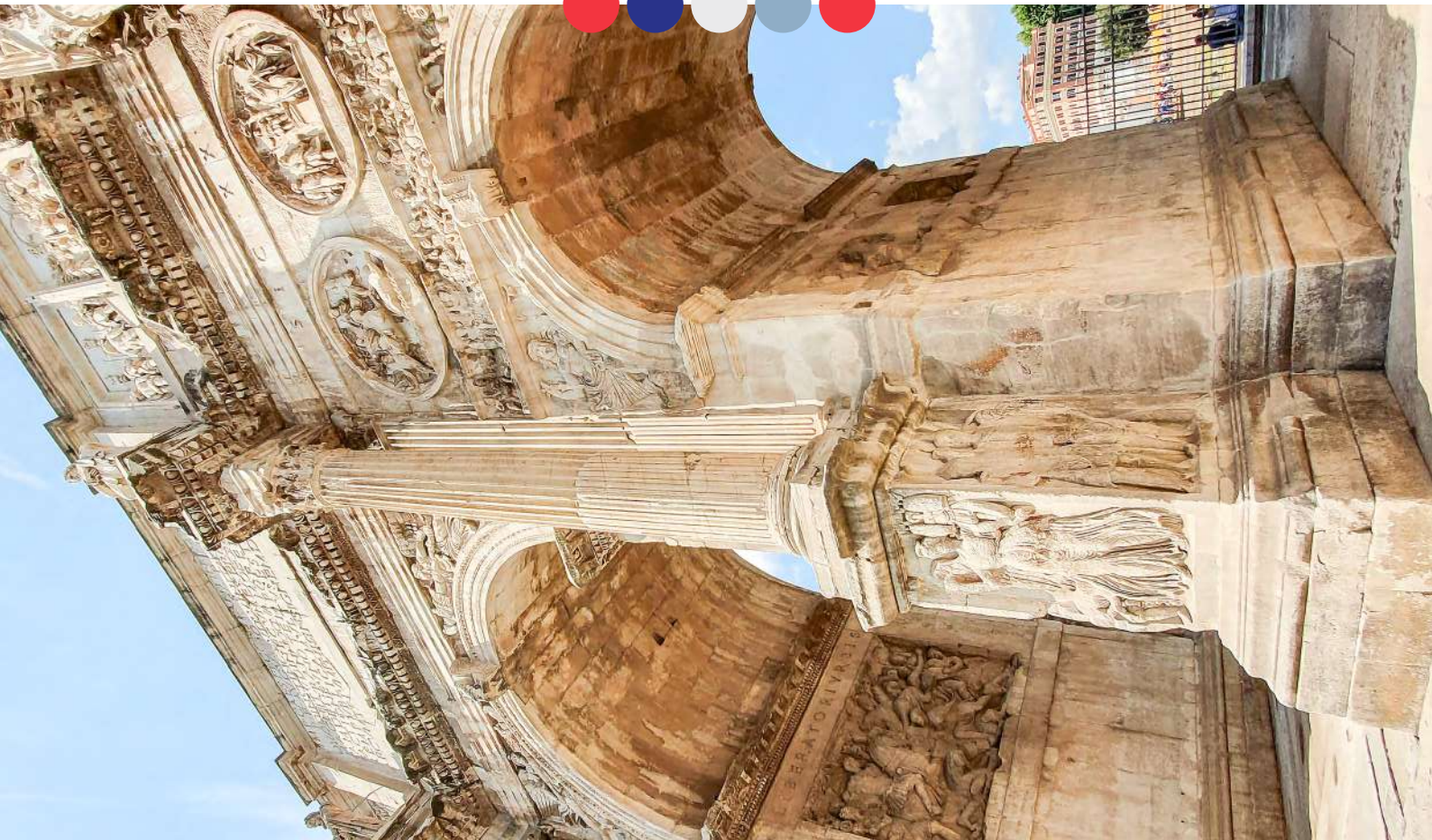
Chairperson: *Giacomo Pietro Comi (Milano), Federica Ricci (Torino)*

Intervengono:

Francesco Ieva	***AltroDomani e Consulta Malattie Neuromuscolari***
Elisabetta Conti	***AIG***
Daniela Lauro	***Famiglie SMA***
Beatrice Vola	***GFB Gruppo Familiari Beta-sarcoglicanopatie***
Paula Morandi	***Mitocon***
Fernanda De Angelis	***Parent Project***
Danila Baldessarri	***Telethon***
Stefania Pedroni	***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 QUETTI[1], LUCA SALI[1], MATILDE CONTARDO[2], ROBERTA DE GIOIA[1], MONICA NIZZARDO[1], ALBERTO ROMANO[1], LORENZO BRAMBILLA[1], LINDA OTTOBONI[2], FEDERICA RIZZO[2], 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 GOGLIA[1], LAURA BOFFA[1], ERICA FREZZA[1], GIULIA GRECO[1], FRANCESCO GRUOSSO[1], ILARIA PETITTA[1], GIULIA NARDINO[1], GIOVANNI VIETRI[1], MARZIA NUCCETELLI[2], ANTONINO DE LORENZO[3], ROBERTO MASSA[1]

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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 BEATRICE[1], ANDREA D'ANGELO[1], JESSICA ONGARO[2], PAOLA RINCHETTI[1], IRENE FARAVELLI[1], MATTEO MIOTTO[3], SIMONA LODATO[3], GIACOMO COMI[1,2], LINDA OTTOBONI[1], 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 ANTONACI[1], MARIKA PANE[1], GIULIA STANCA[1], GIORGIA CORATTI[1], ADELE D'AMICO[2], VALERIA SANSONE[3], BEATRICE BERTI[1], LAVINIA FANELLI[1], EMILIO ALBAMONTE[1], CAROLINA AUSILI[1], ANTONELLA CERCHIARI[2], MICHELA CATTERUCCIA[2], ROBERTO DE SANCTIS[1], DANIELA LEONE[1], CONCETTA PALERMO[1], BIANCA BUCHIGNANI[1], ROBERTA ONESIMO[1], MICHELE TOSI[2], MARIA CARMELA PERA[1], CHIARA BRAVETTI[1], ENRICO BERTINI[2], EUGENIO MERCURI[1]

1. Fondazione Policlinico Gemelli IRCSS; 2. Ospedale Pediatrico Bambino Gesu Ircss;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 CHIAPPINI[1], GIULIA RICCI[1], FRANCESCA TORRI[1], ALESSANDRA GOVONI[1], LAURA MANCA[2], GABRIELE VADI[1], STEFANO ROCCELLA[3], FRANCESCA MAGRI[4], MEGI MENERI[5], FEDERICA FASSINI[4], VERIA VACCHIANO[6], SILVIA TOMASSINI[5], NOEMI GIRONELLA[5], MICHELA COCCIA[5], GIACOMO COMI[4], ROCCO LIGUORI[6], GABRIELE SICILIANO[1]

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 GUERRA[1], ALBERTO MARINI[1], VITTORIA PAGLIARINI[1], CONSUELO PITOLLI[1], GIORGIA CORATTI[1,2], DAVIDE BONVISSUTO[1], CHIARA BRAVETTI[1,2], MARIKA PANE[1,2], EUGENIO MERCURI[1,2], CLAUDIO SETTE[1], MARIA CARMELA PERA[1,2]
1. Universita' Catttolica del Sacro Cuore, Rome, Italy; 2. IRCCS Gemelli, Rome, Italy

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

FRANCESCA DRAGONI[1], BARTOLO RIZZO[1,2], IRENE MARIA DAINESI[3,4], ROSALINDA DI GERLANDO[1,2], STEFANO PARRAVICINI[3,4], ANGELA LUCIA BERARDINELLI[4], STELLA GAGLIARDI[1]
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P08 - A case of Spinal Muscular Atrophy with genetic susceptibility for thrombotic microangiopathies

FEDERICA RICCI[1], ANNA SALVALAGGIO[1], ELEONORA GUARNONE[1], ILARIA CAVALLINA[1], FRANCESCA ROSSI[1], ENRICA ROLLE[1], ROSSELLA D'ALESSANDRO[1], TIZIANA MONGINI[2]
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 RICCI[1], ALESSANDRA SOMÀ[1], ELEONORA GUARNONE[1], ANDREA BUSCAGLIA[1], ROSSELLA D'ALESSANDRO[1], ILARIA CAVALLINA[1], ENRICA ROLLE[1], TIZIANA MONGINI[2], EUGENIO MERCURI[3,39], MARIKA PANE[3,39], GIORGIA CORATTI[3,39], MARTINA RICCI[3,39], ANNA CAPASSO[3,39], ADELE D'AMICO[4], VALERIA SANSONE[5], CLAUDIO BRUNO[6], SONIA MESSINA[7], MICHELA COCCIA[8], GABRIELE SICILIANO[9], ELENA PEGORARO[10], MARA TURRI[11], MASSIMILIANO FILOSTO[12,40], GIACOMO COMI[13,41], RICCARDO MASSON[14], LORENZO MAGGI[15], IRENE BRUNO[16], MARIA GRAZIA D'ANGELO[17], ANTONIO TRABACCA[18], VERA VACCHIANO[19], MARIA DONATI[20], ISABELLA SIMONE[21], LUCIA RUGGIERO[22], ANTONIO VARONE[23], LORENZO VERRIELLO[24], ANGELA BERARDINELLI[25], CATERINA AGOSTO[26], ANTONELLA PINI[27], MARIA ANTONIETTA MAIOLI[28], LUIGIA PASSAMANO[29], FILIPPO BRIGHINA[30], NICOLA CARBONI[31], MATTEO GARIBALDI[32], RICCARDO ZUCCARINO[33], DELIO GAGLIARDI[34], SABRINA SILIQUINI[35], STEFANO PREVITALI[36], DOMENICA TARUSCIO[37], STEFANIA BOCCIA[38], MARIA CARMELA PERA[3,39]
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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 IRCSS, Rome, Italy

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

MARIA IRENE DAINESI[1], ALICE GARDANI[2], CARLO ALBERTO QUARANTA[1], ARIANNA IOSCA[1], SARA FUSCO[1], LAURA CARRARO[1], STEFANO PARRAVICINI[1,2], ANGELA LUCIA BERARDINELLI[2]
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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 LERARIO[1], ELENA ABATI[1], DANIELE VELARDO[1], GIACOMO P. COMI[2], STEFANIA CORTI[1], MONICA SCIACCO[1]

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 OLIVA[1], GABRIELE GIANNOTTA[1], DONATELLA DE GIOVANNI[2], MARTA RUGGIERO[1], IVANA GALLO[1], ISABELLA FANIZZA[1], RITA FISCHETTO[2], ANTONIO TRABACCA[1]

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 GALLO[1], TIZIANA MONGINI[2], FEDERICA RICCI[1], ANTONIO TRABACCA[3], MARIA CARMELA OLIVA[3], GIANCARLO PARENTI[4], SIMONA FECAROTTA[4], SABRINA SILIQUINI[5], FEDERICA DEODATO[6], BENEDETTA GRECO[6], ALESSIA GRAZIOSI[6], SERENA GASPERINI[7], VIOLA CRESCITELLI[7], GAIA KULLMANN[7], RITA MARIA ELISA BARONE[8], LARA CIRNIGLIARO[8], FRANCESCA MENNI[9], FRANCESCA FURLAN[9], ANDREA CELATO[9], FEDERICO MONTI[9], ANGELA BERARDINELLI[10], ARIANNA IOSCA[10], ALICE GARDANI[10], ALBERTO BURLINA[11], VINCENZA GRAGNANIELLO[11], DANIELA GUERALDI[11], CHIARA CAZZORLA[11], ELENA PROCOPIO[12], MICHELE SACCHINI[12]

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P15 - *Pompe Disease: a treatable condition still underdiagnosed*

CARLOTTA POZZI[1], BENEDETTA FRANCESCA RATTI DI DESIO LEVI[2], VALENTINA ROVELLI[3]
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 report

SARA LOPRIENO[1], FRANCESCA TORRI[1], GABRIELE VADI[1], BEATRICE CIURLI[1], FULVIA BALDINOTTI[2], MARIA ADELAIDE CALIGO[2], GRETA ALI'[3], GIOVANNA CENACCHI[4], GIULIA RICCI[1], GABRIELE SICILIANO[1]
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P17 - A novel patient presenting late-onset Polyglucosan Body myopathy due to GYG1 mutations: a case report and review of the literature

NICOLA MOLITIERNO[1], DANIELE VELARDO[2], ELENA ABATI[1,3], SARA ANTOGNOZZI[3], MICHELA RIPOLONE[2], SIMONA ZANOTTI[2], LAURA NAPOLI[2], PATRIZIA CISCATO[2], MONICA SCIACCO[2], GIACOMO PIETRO COMI[1,3], STEFANIA PAOLA CORTI[1,2], DARIO RONCHI[1,3]
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P.18 - Clinical and genetic features of a cohort of patients with myoadenilate deaminase deficiency: a new mutation

CLAUDIA ALBERTI[1], SABRINA LUCCHIARI[1], MARTINA RIMOLDI[2], FRANCESCO FORTUNATO[2], DANIELE VELARDO[1], LAURA NAPOLI[3], 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 review

GABRIELE VADI[1], DOMENIKO HOXHAJ[1], LORENZO BIANCHI[2], LORENZO FONTANELLI[1], FRANCESCA TORRI[1], GIULIA RICCI[1], GABRIELE SICILIANO[1]
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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 USBERGO[1], CARMELO RODOLICO1,2, ANTONIO TOSCANO[1], OLIMPIA MUSUMECI[1,2]
1. Clinical and Experimental Medicine Department, University of Messina, Messina, Italy; 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 URBANO[1], LILIANA VERCELLI[1], MARCO VERCELLINO[2], CHIARA BOSA[3], GIULIO GADALETA[1], GIORGIA BRODINI[1], PAOLA CAVALLA[2], FEDERICA RICCI[4], TIZIANA MONGINI[1]
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P22 - A complex presentation of mitochondrial myopathy
ANNA RUSSO[1], DARIO ZOPPI[1], EMANUELE CASSANO[1], FLORIANA VITALE[1], ROSARIO RUSSO[1], DENISE CASSANDRINI[2], CHIARA NESTI[2], ROSA IODICE[1], FIORE MANGANELLI[1], FILIPPO MARIA SANTORELLI[2], LUCIA RUGGIERO[1]
Department of Neurosciences, Reproductive dnd 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 MARCHET, 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 DIODATO[1], MICHELE TOSI[1], MICHELA CATTERUCCIA[1], FRANCESCO NICITA[1], MARGHERITA VERARDO[1], FRANCESCA CUMBO[1], LUCA BOSCO[2], FABIANA FATTORI[2], ALESSANDRA TORRACO[2], ROSALBA CARROZZO[2], LORENA TRAVAGLINI[2], ENRICO BERTINI[1], ADELE D'AMICO[1]
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 FRANGIAMORE[1], ELENA RINALDI[1], FIAMMETTA VANOLI[1], SILVIA BONANNO[1], FRANCESCA ANDREETTA[1], LORENZO MAGGI[1], ALESSANDRO PINNA[2], ROBERTO ARNABOLDI[2], CARLO ANTOZZI[1], RENATO MANTEGAZZA[1]
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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 VU[1], LAURA FIONDA[2], JAMES F. HOWARD[3], DENIS KOROBKO[4], MAREK SMILOWSKI[5], FIEN GISTELINCK[6], SOPHIE STEELAND[6], JAN NOUKENS[7], JANA PODHORNA[6], YUEBING LI[8], KIMIAKI UTSUGISAWA[9], FRANCESCO SACCÀ[10], HEINZ WIENDL[11], JAN L. DE BLEECKER[12], RENATO MANTEGAZZA[13]
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P29 - A rare case of a late-onset congenital myasthenic syndrome associated to co-occurence of variants in the LRP4 and CHRND genes.
ALESSIA PERNA[1], FILIPPO MARIA SANTORELLI[2], ELISA COLAIZZO[3], SILVIA LA CESA[1], VITTORIO RISO[1], LUDOVICO LISPI[1], ANTONIO PETRUCCI[1]
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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

SOFIA MARINI[1], SILVIA FALSO[1], MARTINA MARINI[1], RAFFAELE IORIO[2]
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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 ANTOZZI[2]
TUAN VU[1], RENATO MANTEGAZZA[2], DJILLALI ANNANE[3], MASAHISA KATSUNO[4], ANDREAS MEISEL[5], MICHAEL NICOLLE[6], VERA BRIL[7], RASHA AGUZZI[8], GLEN FRICK[8], JAMES HOWARD JR[9], CHAMPION MG STUDY GROUP
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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 GROSSKREUTZ[2], ALI AAMER HABIB[3], RENATO MANTEGAZZA[4], JOHN VISSING[5], TUAN VU[6], MARION BOEHNLEIN[7], BERNHARD GREVE[7], FIONA GRIMSON[8], ASHA HAREENDRAN[8], VERA BRIL[9]
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5. Department of Neurology, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark; 6. Department of Neurology, University of South Florida Morsani College of Medicine, Tampa, 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 MARANDO[1], STEFANIA MORINO[1], LUCA LEONARDI[1], LAURA TUFANO[2], ANTONIO LAULETTA[2], ELENA ROSSINI[1], FRANCESCA FORCINA[1], MATTEO GARIBALDI[1,2], GIOVANNI ANTONINI[2], LAURA FIONDA[1]
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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 SORRENTI[1], CAMILLA MIRELLA MARIA STRANO[1], CHRISTIAN LAURINI[1], ADELE RATTI[1], LUCA BOSCO[2], MASSIMO FILIPPI[3], YURI MATTEO FALZONE[1], STEFANO CARLO PREVITALI[4]

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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 FALSO[1], PIETRO ZARA[2], SOFIA MARINI[1], MARIANGELA PUCI[3], ELEONORA SABATELLI[4], GIOVANNI SOTGIU[3], MARTINA MARINI[1], GREGORIO SPAGNI[5], AMELIA EVOLI[1], PAOLO SOLLA[2], RAFFAELE IORIO[4], ELIA SECHI[2]

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

ANNA ROSA AVALLONE[1], LILIANA BEVILACQUA[1], PAOLO ALONGE[2], ANTONINO LUPICA[2], SIMONA MACCORA[2], SONIA AMABILE[3], FILIPPO BRIGHINA[2], VINCENZO DI STEFANO[2], CLAUDIA VINCIGUERRA[1]

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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, Universita' Cattolica del Sacro Cuore, Roma, Italy

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

PIETRO BUSI[1], STEFANO MASCIOCCHI[1], SILVIA SCARANZIN[2], CHIARA MORANDI[2], ANTONIO MALVASO[1], MATTEO SCUCCHI[1], ELISABETTA ZARDINI[2], DIEGO FRANCIOTTA[2], MATTEO GASTALDI[2]

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

SOFIA MARINI[1], MIRIAM FREIMER[2], CHANNA HEWAMADDUMA[3,9], MARIA ISABEL DA SILVA LEITE[4], RAPHAËLLE BEAU LEJDSTROM[5], BABAK BOROOJERDI[6], FIONA GRIMSON[7], NATASA SAVIC[5], JAMES FRANCIS HOWARD JR[8]

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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
Department of Biomedicine, Neuroscience, and Advanced Diagnostics (BIND), University of Palermo, Palermo, Italy

P43 - Kaposi's sarcoma in Myasthenia Gravis patient in oral steroids treatment: two case reports
ADELE BARBACCIA[1], ALESSIA PUGLIESE[1], FIAMMETTA BIASINI[1], ALBA MIGLIORATO[2], CARMELO RODOLICO[1]
1.Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy; 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 CAMERA[1], PAOLO PAONE[1], EMANUELA AGAZZI[1], PAOLA BAZZI[2], DARIO ALIMONTI[1], MANLIO SGARZI[1]
1. ASST Papa Giovanni XXIII, Bergamo, Italy;2.ASST Brianza, Ospedale Vimercate, Vimercate, Italy

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 BARP[1], ISABELLA MORONI[2], MARCELLA NERI[3], FERNANDA FORTUNATO[3], ILARIA VIVALDI3, RICCARDO ZUCCARINO[1], LORENZO MAGGI[4], ELIANA IANNIBELLI[4], MATTEO DE IORIO[5], MARIA IASCONE[6], SILVIA MAZZOLA[7], FRANCESCA GUALANDI[3]
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P.50 - A novel variant in the HSPB3 gene presenting as congenital myopathy. A case report
MICHELE TOSI[1], MARGHERITA VERARDO[1], LUCA BOSCO[2,3], FABIANA FATTORI[3], SERENA CARRA[4], ALESSANDRO ROSA[5,7], MARCO CIRILLO[6], ADELE D'AMICO[1], ENRICO BERTINI[1]
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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 ZANOTTI[1], DARIO RONCHI[2,3], LAURA NAPOLI[1], MICHELA RIPOLONE[1], PATRIZIA CISCATO[1], ROBERTO DEL BO[3], FRANCESCA MAGRI[3], GIACOMO PIETRO COMI[2,3], STEFANIA CORTI[1,2], MONICA SCIACCO[1]
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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 MOLITIERNO[1], DANIELE VELARDO[2], ELENA ABATI[1,3], SARA ANTOGNOZZI[3], MICHELA RIPOLONE[2], SIMONA ZANOTTI[2], LAURA NAPOLI[2], PATRIZIA CISCATO[2], MONICA SCIACCO[2], 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 GAMBELLA[5], LUISA VILLA[5], MARCO RABUFFETTI[6], ALBERTO MARZEGA[6], VALERIO TROMBETTA[1], MARTA CHELI[4], AURORA PARROTTA[5], ENRICA ROLLE[2], ELEONORA TORCHIA[7], BEATRICE RAVERA[7], MAURO MONFORTE[1], JEAN-YVES HOGREL[8], ENZO RICCI[1,7], TIZIANA ENRICA MONGINI[2], SABRINA SACCONI[5], LORENZO MAGGI[4], GIORGIO TASCA[9]

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P55 - SEPN1-related myopathy with acute respiratory onset in middle age: a clinical and genetic study

BARBARA RISI [1], FILOMENA CARIA [1], SIMONA DAMIOLI [1], BEATRICE LABELLA [2,4], ENRICA BERTELLA [1], GIORGIA GIOVANNELLI [1], LUCIA FERULLO [2], EMANUELE OLIVERI [2], MATTIA BUGATTI [3], CARLA BARONCHELLI [3], LORIS POLI [4], 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 BOSCO[1], ADELE RATTI[1], CHRISTIAN LAURINI[1], BENEDETTA SORRENTI[1], CAMILLA MIRELLA MARIA STRANO[1], VALENTINA CANTI[2], PATRIZIA ROVERE QUERINI[2], ANTONIO LAULETTA[3], MATTEO GARIBALDI[3], ALEX VICINO[4], LORENZO MAGGI[4], VINCENZO CARLOMAGNO[5], MATTEO LUCCHINI[5], MASSIMILIANO MIRABELLA[5], MASSIMO FILIPPI[1], UBALDO DEL CARRO[6], STEFANO CARLO PREVITALI[1]

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P57 - RYR1- related myopathies: clinical clues and outcome of a paediatric cohort
ADELE D'AMICO[1], MICHELA CATTERUCCIA[1], DARIA DIODATO[1], IRENE MIZZONI[1], GIACOMO DE LUCA[1], ADELINA CARLESI[2], FABIANA FATTORI[3], LUCA BOSCO[3], MICHELE TOSI[1], ENRICO BERTINI[1]
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P58 - MYH7-related myopathy: an Italian family with atypical phenotype
DOMENICA ZAINO[1], MELISSA BELLINI[2], PAOLO IMMOVILLI[1], ROBERTO PARISI[3], NICOLA MORELLI[4], GIACOMO BIASUCCI[2,5], CHIARA TERRACCIANO[1]
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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'ALESSANDRO[1], ELISA PANERO[1], ILARIA CAVALLINA[1], ENRICA ROLLE[1], FRANCESCA ROSSI[1], FRANCESCA RE[1], FRANCESCA SERTORI[1], LAURA GASTALDI[2], TIZIANA MONGINI[3], FEDERICA RICCI[1]

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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

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

BARTOLO RIZZO[1,2], FRANCESCA DRAGONI[2], MARIA IRENE DAINESI[3,5], ROSALINDA DI GERLANDO[1,5], STEFANO PARRAVICINI[3,5], ANGELA LUCIA BERARDINELLI[4], STELLA GAGLIARDI[5]

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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 GARDANI[1], VALERIA VACCHINI[1], MARIA IRENE DAINESI[2], ARIANNA IOSCA[2], SARA FUSCO[2], STEFANO PARRAVICINI[1], ANGELA LUCIA BERARDINELLI[1]

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

ARIANNA IOSCA[1], ALICE GARDANI[2], SARA FUSCO[1], MARIA IRENE DAINESI[1], LAURA CARRARO[1], CARLO ALBERTO QUARANTA[1], STEFANO PARRAVICINI[1,2], ANGELA LUCIA BERARDINELLI[2]

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

MARIANELA SCHIAVA[1], ROBERT MUNI LOFRA[1], JOHN BOURKE[1], JORDI DÍAZ-MANERA[1], MEREDITH JAMES[1], MAHA ELSEED[1], MONIKA MALINOVA[1], JASSI MICHEL-SODHI[1], DIONNE MOAT[1], ELISABETTA GHIMENTON[1], MICHELLE MCCALLUM[1], CARLA FLORENCIA BOLAÑO DÍAZ[1], ANNA MAYHEW[1], KAREN WONG[1], MARK RICHARDSON[1], GIORGIO TASCA[1], GAIL EGLON[1], MICHELLE EAGLE[2], CATHY TURNER[1], EMMA HESLOP[1],

VOLKER STRAUB[1], CHIARA MARINI BETTOLO[1], MICHELA GUGLIERI[1]
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P66 - MITRACLIP implantation in a patient with Duchenne Muscular Dystrophy

CONCETTA PALERMO[1], DANIELA LEONE[1], BIANCA BUCHIGNANI[1], PRISCILLA LAMENDOLA[2], 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 FUSCO[1], STEFANO PARRAVICINI[2], ARIANNA IOSCA[1], MARIA IRENE DAINESI[1], LAURA CARRARO[1], CARLO ALBERTO QUARANTA[1], MICHELE TERZAGHI[3], ANGELA BERARDINELLI[2]
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P68 - The DMD Hub Central Recruitment Database, a model for effective recruitment to Rare Disease clinical trials

Presented by: PIETRO RIGUZZI [2]
MICHELA GUGLIERI[1], PHILLIP CAMMISH[1], PIETRO RIGUZZI[1,2], ALEX JOHNSON[3], KERRY SHIPPEY[3], KATE ADCOCK[4], SUZANNE GLOVER[5], EMILY REUBEN[3], MARIA ELENA FARRUGIA[6], ROSALINE QUINLIVAN[7], TRACEY WILLIS[8], ANNE-MARIE CHILDS[9], MARIACRISTINA SCOTO[10], CATHY TURNER[1], MEGAN MCNIFF[1], DAN RISEBOROUGH[1], VOLKER STRAUB[1], EMMA HESLOP[1]
1. John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, Newcastle Upon Tyne, UK;2. Department of Neurosciences, University of Padova, Padova, Italy;3. Duchenne UK;4. Muscular Dystrophy UK; 5. Pathfinders Neuromuscular Alliance, UK; 6. Institute of Neurological Sciences, Glasgow, Scotland, UK;7. Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology and The National Hospital for Neurology and Neurosurgery, London, UK; 8.Wolfson Centre for Inherited Neuromuscular Diseases and Department of Musculoskeletal Histopathology, Rjah Orthopaedic Hospital, Oswestry, UK;9.Leeds Teaching Hospitals NHS Trust, London, UK; 10. Great Ormond Street Hospital NHS Trust, London, UK

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 VERA [1], RAOUL ROOMAN [1], ERIC P HOFFMAN [2], PAULA R CLEMENS [3], MICHELA GUGLIERI [4], 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 HENRICSON,[1] ANA DE VERA,[2] MIKA LEINONEN,[2] PAULA R CLEMENS,[3] MICHELA GUGLIERI,[4] NATALIE TRUBA,[5] ERIC P HOFFMAN[6]
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P73 - Givinostat in DMD: results of the Epidys study with particular attention to NSAA

EUGENIO M. MERCURI[1], CLAUDIA BROGNA[2], MAH JEAN[3], NATHALIE GOEMANS[4], ERIC NIKS[5], SARA CAZZANIGA S[6], NICOLETTA COCEANI[4], PAOLO BETTICA P[6], CRAIG MCDONALD[7]

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

KRISTA VANDENBORNE[1], REBECCA WILLCOCKS[1], GLENN WALTER[2], SEAN FORBES[3], SARA CAZZANIGA[4], PAOLO BETTICA[4], EUGENIO M. MERCURI[5], CRAIG MCDONALD[6]

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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 MARINELLI[1], PAOLA MANTUANO[1], ORNELLA CAPPELLARI[1], BRIGIDA BOCCANEGRA[1], LISAMAURA TULIMIERO[1], ANTONIETTA MELE[1], DANIELA TRISCIUZZI[1], ENRICA CRISTIANO[1], ELENA CONTE[1], ELENA BRESCIANI[2], ANTONIO TORSELLO[2], SEVERINE DENOYELLE[3], ORAZIO NICOLOTTI[1], ANTONELLA LIANTONIO[1], ANNAMARIA DE LUCA[1]

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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 PANE [1], GUILLAUME BASSEZ [2], JORDI DIAZ-MANERA [3], JAMES B. LILLEKER [4], BAZIEL VAN ENGELEN [5], RICHARD H ROXBURGH [6], BENEDIKT SCHOSER [7], CHRISTOPHER TURNER [8], CHRIS MIX [9], SOMA RAY [9], BAOGUANG HAN [9], WILDON FARWELL [9], DANIEL WOLF [9], ERWAN DELAGE [9], VALERIA SANSONE [10]

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

SALVATORE ROSSI[1,2], ANTONIO FUNCIS[2], GIANMARCO DALLA ZANNA[2], AURELIA ZAULI[3], 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 RISI[1], ANDREA PILOTTO[2,3,4], ANDREA RIZZARDI[2,4], ELISABETTA FERRARI[1], BEATRICE LABELLA[2,4], CINZIA ZATTI[2,4], CLINT HANSEN[5], ROBBIN ROMIJNDERS[5], FILOMENA CARIA[1], SIMONA DAMIOLI[1], ENRICA BERTELLA[1], LORIS POLI[4], LUCIA FERULLO[2,4], EMANUELE OLIVIERI[2,4], WALTER MAETZLER[5], 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 ALTAMURA[1], CARMEN CAMPANALE[1], PAOLA LAGHETTI[1], ILARIA SALTARELLA[1], DAMIEN STERNBERG[2], SAVINE VICART[2], JEAN-FRANCOIS DESAPHY[1]

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P80 - Irisin and brain-derived neurotrophic factor in Myotonic Dystrophy type 1
ERICA FREZZA[1], MARIANGELA GOGLIA[1], GIULIA GRECO[1], VIETRI GIOVANNI[1], ILARIA PETITTA[1], FRANCESCO GRUOSSO[1], GIULIA NARDINO[1], LAURA BOFFA[1], MARZIA NUCCETELLI[2], ROBERTO MASSA[1]
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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 IACOBELLI[1], MARTINA RIMOLDI[2], SABRINA LUCCHIARI[1], SERENA PAGLIARANI[2], GRAZIA D'ANGELO[3], ROCCO LIGUORI[4,14], FRANCESCO OTTAVIO LOGULLO[5], ELENA PEGORARO[6], VALERIA SANSONE[7], LUCIO SANTORO[8], MARINA SCARLATO[9], GABRIELE SICILIANO[10], GABRIELLA SILVESTRI[11], GIUSEPPE VITA[12], 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 CROCE[1], FRANCESCA VALENTINO[2], EMANUELE MICAGLIO[3], SARA BENEDETTI[3], MATTEO PAOLETTI[2], GIUSEPPE COSENTINO[2], SABRINA RAVAGLIA[2]
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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 COLACICCO[1], CAROLA RITA FERRARI AGGRADI[1], MANACORDA FILIPPO[2], RICCARDI SEBASTIANO[2], CASIRAGHI JACOPO[1], BARP ANDREA[3], STANO SALVATORE[3],

LIZIO ANDREA[1], VITALI PAOLO[4], ZANARDO MORENO[4], SARDANELLI FRANCESCO[4], ZUCCARINO RICCARDO[3], SANSONE VALERIA[1]
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P85 - Psoriasis and Myotonic Dystrophy type 1: another cutaneous manifestation of a multisystemic disorder

LAURA TUFANO[1], ELISABETTA BUCCI[2], GIOVANNI ANTONINI[2], MATTEO GARIBALDI[1]
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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 TRAVERSO[1], MICHELE IACOMINO[2], SERENA BARATTO[3], ELENA FAEDO[4], ALESSANDRO GEROLDI[4], PAOLO UVA[5], 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 VOLA[1], MASSIMILIANO CERLETTI[2], CARLES SANCHEZ RIERA[3], OREST SEMERYAK [4], CHAHNEZ CHARFI TRIKI[5], YVAN TORRENTE[6]

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P88 - IPSCs generation, characterization and differentiation for LGMDR4 patients

YVAN TORRENTE[1], GIULIO POMPILIO[2], MASSIMILIANO CERLETTI[3], CARLES SANCHEZ RIERA[4], FRANÇOIS GROS-LOUIS[5], BEATRICE VOLA[6]

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

MICHELA CATTERUCCIA[1], DARIA DIODATO[1], FABIANA FATTORI[2], MARCO SAVARESE[3], LUCA BOSCO[1,2], ENRICO BERTINI[1], ADELE D'AMICO[1]

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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 PENZO[1], GIORGIA SEGRE[1], SARA GIBERTINI[2], LORENZO MAGGI[2], GIOVANNA ZORZI[1], ISABELLA MORONI[1]
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P92 - Clinical characterization of a large monocentre cohort of patients with Becker Muscular Dystrophy
PIETRO RIGUZZI[1,2], HOLLY BORLAND[1], JOHN BOURKE[1], CHIARA MARINI BETTOLO[1], MEREDITH JAMES[1], ROBERT MUNI LOFRA[1], JORDI DIAZ-MANERA[1], GIORGIO TASCA[1], MAHA ELSEED[1], MARIANELA SCHIAVA[1], CARLA BOLANO DIAZ[1], JASSI MICHEL-SODHI[1], DIONNE MOAT[1], KAREN WONG[1], ELENA PEGORARO[2], LUCA BELLO[2], VOLKER STRAUB[1], MICHELA GUGLIERI[1]
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P93 - A novel de novo SYNE2 mutation causing a childhood form of type 5 Emery-Dreifuss Muscular Dystrophy
ESTHER PICILLO[1], ORLANDO PACIELLO[2], MARIA ELENA ONORE[1], ALBERTO PALLADINO[1], ELIA CESARONE[1], LUIGIA PASSAMANO[1], VINCENZO NIGRO[1], LUISA POLITANO[3]
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P94 - Longitudinal echocardiographic measures in Becker muscular dystrophy
GIULIANA CAPECE[1], ANGELA PETROSINO[1], ELENA SOGUS[1], PIETRO RIGUZZI[1], CHIARA CALORE[2], ELENA PEGORARO[1], LUCA BELLO[1]
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P95 - Subjective assessment of sleep quality in adult patients with dystrophinopathy: a single centre experience.
EMANUELE OLIVIERI[1], LUCIA FERULLO[1], LEANDRO PURIN[1], BEATRICE LABELLA[1], BARBARA RISI[2], FILOMENA CARIA[2], SIMONA DAMIOLI[2], LORIS POLI[3], MARIA PIA PASOLINI[3], ALESSANDRO PADOVANI[1], MASSIMILIANO FILOSTO[1]
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2. Nemo-Brescia Clinical Center for Neuromuscular Diseases, Brescia, Italy;
3. Unit of Neurology, Asst Spedali Civili, Brescia, Italy

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

BEATRICE RAVERA[1],SARA BORTOLANI[2], ELENA TORCHIA[1], MAURO MONFORTE[2], GIORGIO TASCA[3], ENZO RICCI[1,2]

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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 CONTE[1], PAOLA IMBRICI[1], GIORGIA DI NOI[1], KONRAD URBANEK[2], DONATO CAPPETTA[3], LIBERATO BERRINO[4], ANNAMARIA DE LUCA[1], ANTONELLA DE ANGELIS[4], ANTONELLA LIANTONIO[1]

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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 GADALETA[1], LILIANA VERCELLI[1], GUIDO URBANO[1], ENRICA ROLLE[1], LOREDANA CHIADO'-PIAT[1], FEDERICA RICCI[2], TIZIANA MONGINI[1]

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

MARIA FRANCESCA DI FEO[1,28], ALI OGHABIAN[1], ELLA NIPPALA[1], FRANCESCA FORZANO[2], EDOARDO MALFATTI[3], CLAUDIA CASTIGLIONI[4], MATHIAS GAUTEL[5], HEINZ JUNGBLUTH[5], ILONA KREY[6], DAVID GOMEZ ANDRES[7], ANGELA BRADY[8], MARIA IASCONE[9], ANNA CEREDA[10], LIDIA PEZZANI[10], DANIEL NATERA DE BENITO[11], ANDRES NASCIMIENTO OSORIO[12], BERTA ESTÉVEZ ARIAS[13], SERGEI KURBATOV[14], TANIA ATTIE-BITACH[15], SHEELA NAMPOOTHIRI[16], ERIN RYAN[17], MICHELLE MORROW[18], SVETLANA GOROKHOVA[19], BRIGITTE CHABROL[20], JUHA SINISALO[21], HELI TOLPPANEN[21], JOHANNA TOLVA[22], FRANCINA MUNELL[23], JESSICA CAMACHO SORIANO[24], MARIA ANGELES SANCHEZ DURAN[25], MRIDUL JOHARI[26], HOMA TAJSHARGHI[27], PETER HACKMAN[1], BJARNE UDD[1], MARCO SAVARESE[1]

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Neuropaediatrics Department, Hospital Sant Joan De Déu, Institut De Recerca Sant Joan De Déu, Barcelona, Spain;12. Neuropaediatrics Department, Institut de Recerca Pediàtrica Hospital Sant Joan De Déu, Barcelona, Spain; 13. Neuromuscular Unit, Department of Neurology, Hospital Sant Joan De Déu, Barcelona, Spain; 14. Regional Medical Diagnostic Centre, Voronezh, Russia; 15. Unité D'embryofoetopathologie, Service D'histologie-Embryologie-Cytogénétique, Hôpital Necker-Enfants Malades, APHP, Paris, France;16. Department of Pediatric Genetics, Amrita Institute Of Medical Sciences & Research Centre, Kerala, India; 17. Genomic Data / Genetic Counseling, Genedx, Gaithersburg, Maryland, USA; 18. Genedx, Gaithersburg, MD, USA; 19. Marseille Medical Genetics, Aix Marseille Université, Faculté des Sciences Médicales et Paramédicales; 20. APHM, CHU Timone, Centre de Reference des Maladies Neuromusculaires de L'enfant Pacarare, Service de Neuropediatrie, Marseille, France;
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28. University of Genova, Genova, Italy

P101 - Biallelic HMGCS1 variants in rigid spine syndrome

CHIARA FIORILLO[1], MONICA TRAVERSO[1], MARCELLO SCALA[1], SERENA BARATTO[1], CLAUDIO BRUNO[1], MARINA PEDEMONTE[1], FEDERICA TRUCCO[1], FEDERICO ZARA[1], GINA RAVENSCROFT[2]
1. IRCCS Istituto Gaslini, Genova, Italy; 2. Harry Perkins Institute of Medical Research, University of Westerm Australia,Australia

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

MARIANNA FARNÈ[1], FERNANDA FORTUNATO[1], SILVIA MONTANARI[1], ALICE MARGUTTI[1], VITTORIA NAGLIATI[1], RITA SELVATICI[1], CHRISTINA SAIER[2], JAN KIRSCHNER[2], STEFAAN SANSEN[3], SILVIA OTTOMBRINO[4], EMANUELE AGOLINI[4], ANTONIO NOVELLI[4], ENRICO SILVIO BERTINI[5], ALDONA ZYGMUNT[6], ALESSANDRA FERLINI[1]
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P103 - Long-read DNA and RNA sequencing improves analyzing large and complex genes: the example of TTN and NEB

RAFAELA OWUSU[1], ELLA NIPPALA[1], ALI OGHABIAN[1], MARIA FRANCESCA DI FEO[2], MEHARIJI ARUMILLI[1], DAVID GOMEZ ANDRES[3], LYDIA SAGAT[4], VILMA LOTTA LEHTOKARI1, JESSICA CAMACHO SORIANO[3], MARIA ANGELES SANCHEZ DURAN[3], FRANCINA MUNELL[3], KATARINA PELIN[1], PETER HACKMAN[1], BJARNE UDD[1], MARCO SAVARESE[1]
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P104 - Severe epileptic encephalopathy with progressive cerebral and cerebellar atrophy and peripheral neuropathy: a new GEMIN5 phenotype?

DARIA DIODATO[1], GINEVRA ZANNI[1], ALESSANDRA TERRACCIANO[1], PAOLA DE LISO[2], CONCETTA LUISI[2], MARCELLO NICETA[3], MARCO TARTAGLIA[3], ENRICO BERTINI[1], ADELE D'AMICO[1]

1. Muscular And Neurodegenerative Disease Unit; 2. Epilepsy And Movement Disorders Neurology Unit;3. Molecular Genetics, Bambin Gesù Hospital, Rome, Italy

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

MARTA CHELI[1], DENISE CASSANDRINI[2], DANIELE GALATOLO[2], GAETANO VATTEMI[1], FILIPPO MARIA SANTORELLI[2], PAOLA TONIN[1]

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

GIULIA MARCHETTO[1], DENISE CASSANDRINI[2], FRANCESCO DALLA BARBA[3], MARTA CHELI[1], DIEGO LOPERGOLO[4], ALESSANDRO MALANDRINI[4], CARMELO RODOLICO[5], FILIPPO MARIA SANTORELLI[2], PAOLA TONIN[1], MARCELLO CAROTTI[3], DORIANNA SANDONÀ[3], GAETANO VATTEMI[1]

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Others

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

Chairperson: *Maria Sframeli (Messina)*

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

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

CLAUDIA ALBERTI[1], DOMENICA SACCOMANNO[1], ALESSIA ANASTASIA[2], GRAZIA D'ANGELO[1], MARIA TERESA BASSI[1], LORENZO QUETTI[2], LUCA SALI[2], LORENZO BRAMBILLA[2], ALBERTO ROMANO[2], GIACOMO PIETRO COMI[1,2], FEDERICA RIZZO[2], STEFANIA CORTI[1,3], ELENA ABATI[1,2]
1. Department of Pathophysiology and Transplantation (Dept), Dino Ferrari Centre, Neuroscience Section, University of Milan, Milan, Italy; 2. Neurology Unit, Fondazione Irccs Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; 3. Neuromuscular and Rare Diseases Unit, Fondazione Irccs Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy

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 BUSINARO[1], STEFANO MASCIOCCHI[1], SILVIA SCARANZIN[2], CHIARA MORANDI[2], ELISABETTA ZARDINI[1], DIEGO FRANCIOTTA[2], MATTEO GASTALDI[1]
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 BARRESI[1], ANA TOPF[2], ALEJANDRO GONZALEZ-CHAMORRO[2], JORDI DIAZ-MANERA[2], SARAH SHIRA EMMONS[3], LAURA RUFIBACH[3]
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 FIORILLO[1], ROBERTA PIERA BENCIVENGA, CHIARA GRASSO, MARIANGELA IAPOCE, LUCIA RUGGIERO
Department of Neurosciences, Reproductive dnd 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 CORTESE[1], MARIANNA BRIENZA[1], LAURA DE GIGLIO[1], ROSALBA CARROZZO[2], ALESSANDRA TORRACO[2], FABIANA FATTORI[3], LUCA BOSCO[4], MARIA CONCETTA ALTAVISTA[5], ELENA MARIA PENNISI[1]

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

ANGELA LA TEMPA [1,2,7], FEDERICA TRENTIN [1,2,7], GIULIA GUARDI [1,2,7], MELANIA GIANNOTTA [2], FLAVIA PALOMBO [3], AHMED SHEIK MAYE HODMAN [4], LAURA LICCHETTA [5], MARIA LUCIA VALENTINO [5], ROSARIA PLASMATI [6], DUCCIO MARIA CORDELLI [2,7], ANTONELLA PINI [2]
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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 GIANNOTTA [1], ROSARIA PLASMATI [4], FLAVIA PALOMBO [5], DUCCIO MARIA CORDELLI [2,3], ANTONELLA PINI [1]
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P116 - New association between the ZASP/LDB3 Pro26Ser variant and inclusion body myositis.

DANIELA PIGA [1], SIMONA ZANOTTI [2], MICHELA RIPOLONE [2], LAURA NAPOLI [2], PATRIZIA CISCATO [2], SARA GIBERTINI [3], LORENZO MAGGI [3], FRANCESCO FORTUNATO [4], ANDREA RIGAMONTI [5], 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 VENANZI[1], NICCOLÒ CAMPAGNA[1], TICCI CHIARA[2], FERRI LORENZO[3], MORRONE AMELIA[3], SACCHINI MICHELE[2], PROCOPIO ELENA[2]
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P118 - Countermeasures for sarcopenia: effects of BCAA-based formulations on muscle function, histomorphology, and calcium homeostasis in aged mice.

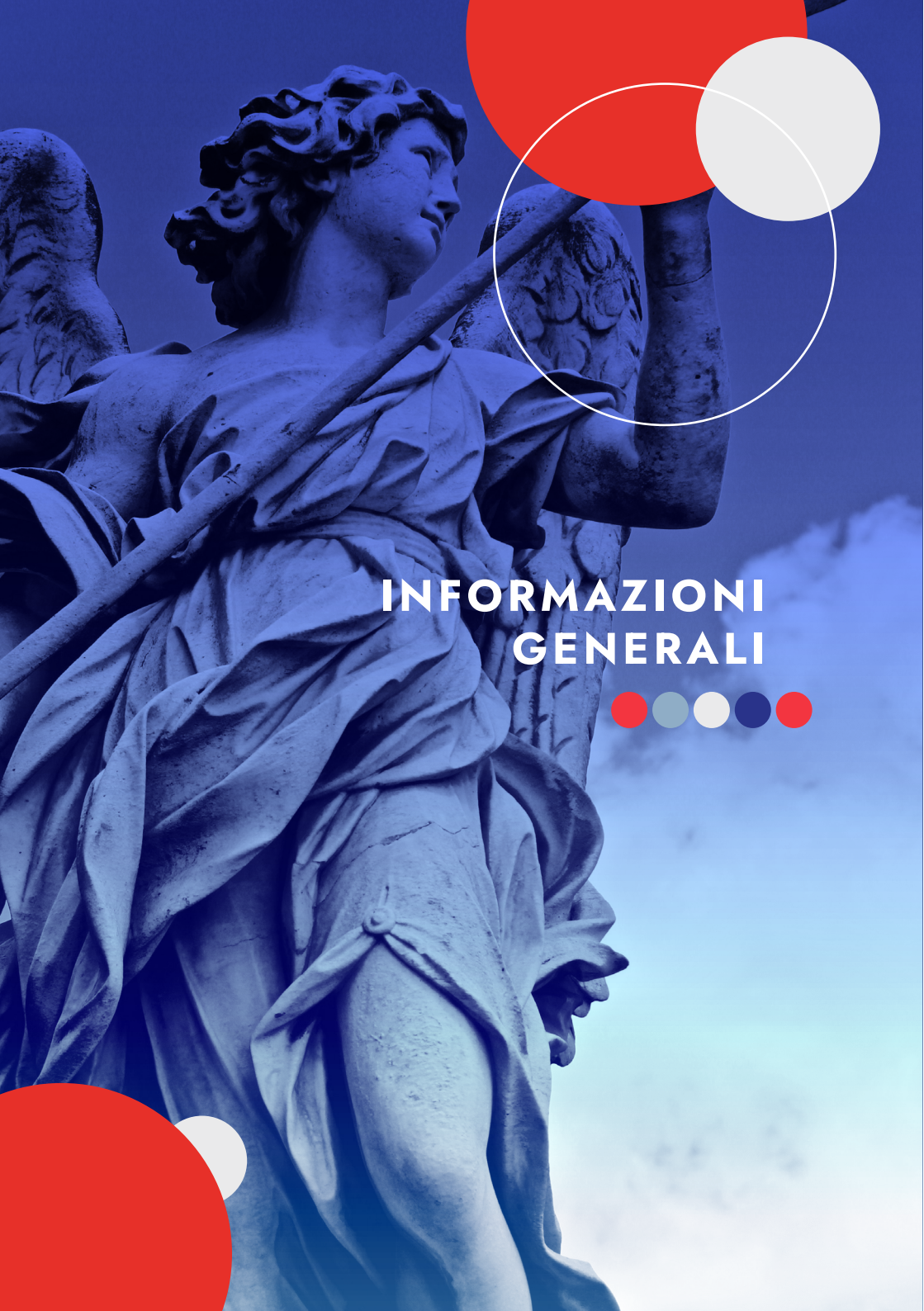
ROBERTA LENTI[1], PAOLA MANTUANO[1], ELENA CONTE[1], BRIGIDA BOCCANEGRA[1], GIANLUCA BIANCHINI[2], ORNELLA CAPPELLARI[1], LISAMAURA TULIMIERO[1], MICHELA DE BELLIS[1], ANTONIETTA MELE[1], ANTONELLA LIANTONIO[1], MARCELLO ALLEGRETTI[2], ANDREA ARAMINI[2], ANNAMARIA DE LUCA[3]

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 DIELLA[1], FABIO STORM[1], LUCA MOLTENI[1], MORENA DELLE FAVE[1], GIULIA CANELLA[1], GIOVANNI MEOLA[2], EMILIA BIFFI[1], MARIA GRAZIA D'ANGELO[1]

1. Scientific Institute Eugenio Medea- Bosisio Parini; 2. University of Milan



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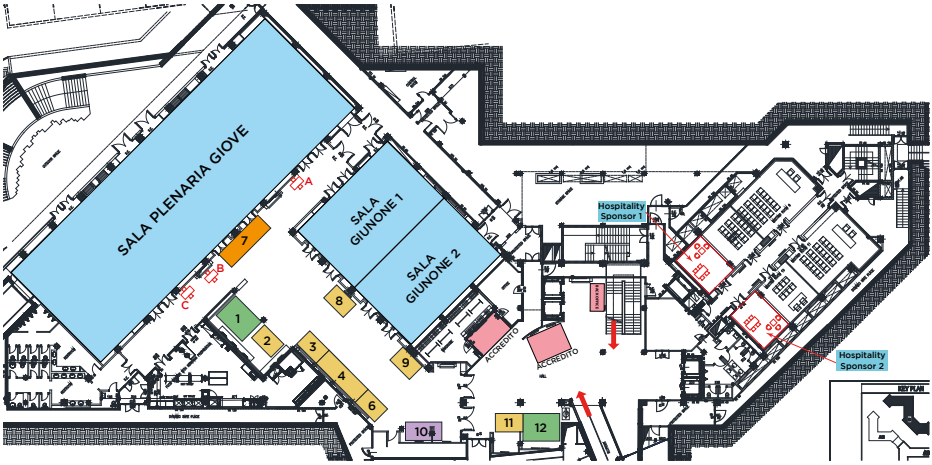

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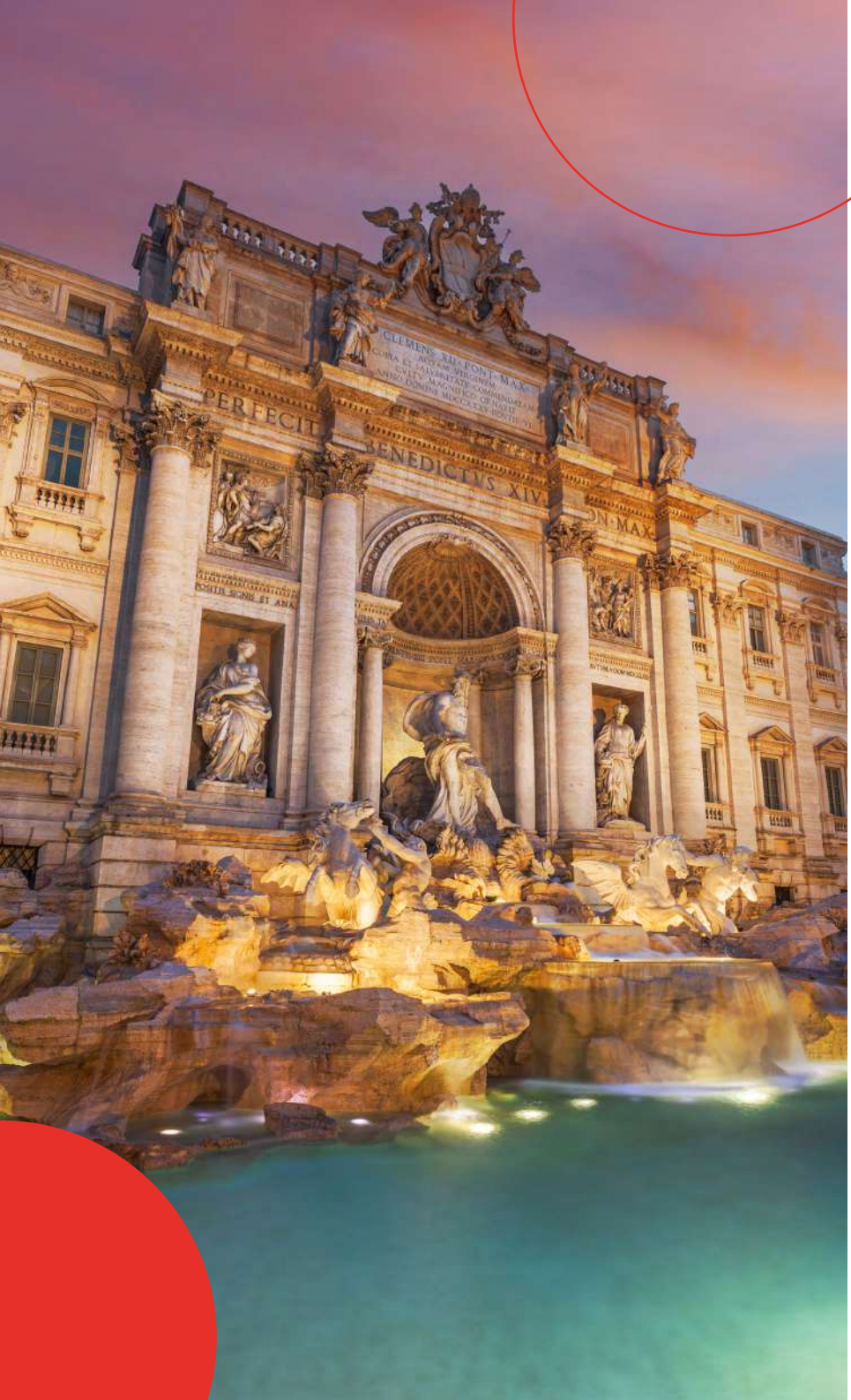
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