XXIV National Congress of Italian Association of Myology (AIM)

Rome, 5-8 June 2024

Wednesday 5 June 2024

PLENARY ROOM

14:30 – 15:00
Chairperson: TBD
Greetings and Introduction | Alfredo Berardelli (Rome)

15:00 – 15:30
Chairpersons: TBD
Lecture 1: The increasing complexity in the diagnoses and treatment of genetic muscle diseases.
| Volker Straub (Newcastle upon Tyne)

15:30 – 17:00
Chairpersons: TBD
Workshop Myotonic Dystrophy- Part I

15:00 Unravelling the complexity of the DM repeat expansions: implications for the genotype-phenotype correlations | Annalisa Botta (Rome)

15:30 Identifying biomarkers of organ involvement in DM1 |Gabriella Silvestri (Rome)

16:00 Towards an Italian consensus on the management of cardiological alterations in the DM | Vincenzo Russo (Naples)

16:30 Ipersonnia e disturbi respiratori sonno-correlati nella distrofia miotonica di tipo 1 e tipo tipo 2: diagnosi e trattamento | Andrea Romigi (Pozzilli)

17:00 – 17:30
COFFEE BREAK

17:30 – 18:00
Workshop Myotonic Dystrophy- Part II
Chairperson: TBD

17:30 Outcome and intervention in Paediatric DM1 | Valeria Sansone (Milan)

17:40 Treatment development pipeline in the DM1 | Roberto Massa (Rome)

17:50 Myotonic dystrophy type 2: still underdiagnosed? How to improve disease awareness | Federica Montagnese (Messina)

18:00 – 18:30
Chairperson: TBD
Lecture 2: Preimplantation genetic testing and prenatal diagnosis: fundamental rights and bioethics | Stefano Canestrari (Boulogne)

18:30 – 19:00
LECTURE – SPONSORIZED BY NOVARTIS - NO CME
The latest evidence in the treatment paradigm of Spinal Muscular Atrophy.
Chairperson: Marika Pane (Rome)

From Clinical Trials to Clinical Practice: anticipating the future and exploring novel aspects of the disease. Adele D'Amico (Rome) and Antonio Varone (Naples)

19.00-21.00 GET TOGETHER COCKTAIL

Thursday 6 June 2024

PLENARY ROOM

08:30 – 09:15
SYMPOSIUM - SPONSORIZED BY DYNE - NO CME
Advancements in Neuromuscular Disorder Therapeutics: Lessons Learned in DMD and Emerging Perspectives in DM1 Therapeutic Development
Chairperson: Eugenio Mercuri (Rome)

08:30 Lessons Learned from DMD and Update on the DELIVER Ongoing Phase 1/2 Trial | Marika Pane (Rome)

08:55 Insights from DM1 Natural History and an Update on the ACHIEVE Ongoing Phase 1/2 Clinical Trial | Valeria Sansone (Milan)

PLENARY ROOM

09:15 - 09:45
Chairperson: TBD
Promoting brain health through novel anti-seizure treatments | Paul Boon (Eindhoven)

09:45 -10:15
Chairperson: TBD
Lecture: Epilepsy in Mitochondrial Disorders | Michelangelo Mancuso (Pisa)

10:15 – 10:30
COFFEE BREAK

10:30 – 11:30
Chairperson: TBD
Muscle TriviAIM - Fase eliminatoria

11:30 – 13:00
PLENARY WORKSHOP 1
Chairperson: TBD
Current and future perspectives of genomic NBS: are we ready to deal with it?

11:30 Where are we now | Jim Bonham (Sheffield)

11:50 Genome NBS: the Screen4Care EU project | Alessandra Ferlini (Ferrara)

12:20 Barriers and opportunities | Vincenzo Nigro (Naples)

12:40 Il lato politico: LEA vs real life | Orfeo Mazzella (Torre Annunziata)

PARALLEL ROOM 1

11:30 – 13:00
POSTER SESSION I - NO CME

13:00 – 14:00
LIGHT LUNCH

PLENARY ROOM

14:00 – 15:00
SYMPOSIUM – SPONSORIZED BY BIOGEN - NO CME

Biomarkers and the Development of an Evidence Based Medicine Approach in Spinal Muscular Atrophy

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

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

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

14:50 Discussion

PLENARY ROOM

15:00 – 16:00
SYMPOSIUM – SPONSORIZED BY ROCHE - NO CME

THE NEW GENERATION OF SMA PATIENTS
Chairperson: Marika Pane (Roma)

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

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

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

15.45 Discussion

16:00–16:30
COFFEE BREAK

PLENARY ROOM

16:30-17:30
Chairperson:TBD
Oral Communication | Myastenia - Transversal

16:30 Ocular myasthenia vs generalized myasthenia gravis with ocular onset: clinical outcomes from a single-centre retrospective study | Alessia Pugliese (Messina)

16:42 Preferential binding to adult or fetal acetylcholine receptor isoform as a promising predictive biomarker in myasthenia gravis | Francesca Beretta (Florence)

16:54 A multicentre, prospective study comparing autoantibody diagnostic assays in myasthenia gravis | Laura Degiglio (Rome)

17:06 Effectiveness of thymectomy in juvenile myasthenia gravis: preliminary results of a15-year follow up study | Michelangelo Maestri Tassoni (Pisa)

17:18 Clinical outcome measures in a prospective cohort of myasthenia gravis patients | Massimiliano Ugo Verza (Florence)

PARALLEL ROOM 1

16:30 – 17:30
Chairperson: TBD
Oral Communication | Myotonic Dystrophies

16:30 Cardiac risk and myocardial fibrosis assessment with Cardiac Magnetic Resonance in patients with Myotonic Dystrophy | Elena Abati (Milan)

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

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

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

17:18 Proof of concept for drug repurposing of fenamates in myotonia congenita | Ilaria Saltarella (Bari)

PARALLEL ROOM 2

16:30 – 17:30
Chairperson: TBD
Oral Communication | Metabolic Myopathies

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

16:42 Quantitative muscle MRI in individual thigh muscles in early Pompe disease
Michele Giovanni Croce (Pavia)

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 (Messina)

17:06 Clinical and laboratory follow-up in a cohort of lipid storage myopathies: a single center experience
Mattia Porcino (Messina)

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 (Brescia)

PLENARY ROOM

17:30 – 18:30
Chairperson: TBD
Oral Communication | Myastenia – Therapies

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 | Rita Frangiamore (Milan)

17:42 Efgartigimod in non-AChR generalized myasthenia gravis | Carlo Antozzi (Milan)

17:54 A Real-life experience with Eculizumab and Efgartigimod in generalized Myasthenia Gravis patients | Laura Fionda (Milan)

18:06 Rituximab in refractory myasthenia gravis: 5 year single center follow up | Dario Ricciardi (Naples)

18:18 Eculizumab in refractory generalized Myasthenia Gravis: a single center 1 year experience
Carmen Erra (Naples)

PARALLEL ROOM 1

17:30 – 18:30
Chairperson: TBD
Oral Communication | Dystrophies

17:30 SYNE-1 and SYNE-2 mutations: expanding the genotype and phenotype spectrum of nesprinopathies Marta Cheli (Verona)

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 (Boulogne)

17:54 In search of biomarkers for Emery-Dreifuss Muscular Dystrophy | Giovanna Lattanzi (Boulogne)

18:06 Novel biomarkers for limb-girdle muscular dystrophy associated to CAPN3 mutation
Diego Lopergolo (Siena)

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 (Pisa)

PARALLEL ROOM 2

17:30 – 18:30
Chairperson: TBD
Oral Communication | Latest Development in Myopathies

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

17:45 Myositis with mitochondrial pathology: a multicentric case series | Antonio Lauletta (Rome)

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

18:15 Treatment of active idiopathic inflammatory myopathies by inhibiting FcRn: Pre-registration report of ALKIVIA, a phase 2/3 trial with efgartigimod | Massimiliano Mirabella (Rome)

18:30 – 19:00
Chairperson: TBD
Lecture 4: State of the art of therapy for Duchenne Muscular Dystrophy |Luca Bello (Padua)

19:00 – 19.30

LECTURE – SPONSORIZED BY ITALFARMACO - NO CME

The Italian contribution to scientific innovation in Duchenne muscolare dystrophy research

Eugenio Mercuri (Rome)

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Friday 7 June 2024

PLENARY ROOM

08:30-10:00
Chairperson: TBD
PLENARY WORKSHOP 2
Sporadic Inclusion Body Myositis: Updates on diagnostic clues and therapeutic perspective

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

09:00 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)

09:30 Looking for reliable clinical and instrumental outcome measures in s-IBM | Matteo Lucchini (Rome)

PARALLEL ROOM 1

08:30 – 10:00
Chairperson: TBD
PARALLEL WORKSHOP 3
Diagnostic tools for congenital myopathies

08:30 Clinical framing | Margherita Milone (Rochester)

08:50 Genomic approach | Marco Savarese (Helsinki)

09:10 MYO-MRI for congenital myopathies | Anna Pichiecchio (Pavia)

09:20 Biomarkers | Ester Zito (Milan)

10:00 – 10:30
Chairperson: TBD
Lecture 5: Diagnosis and treatment in Pompe Disease | Olimpia Musumeci (Messina)

10:30 – 11:00
COFFEE BREAK

PLENARY ROOM

11:00 – 12.30
Chairperson: TBD
PLENARY WORKSHOP 4
Mitochondrial diseases: from the emergency department to the new therapies. Avenues and pitfalls

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

11:20 Update on Leigh Syndrome | Enrico Bertini (Rome)

11:40 Therapeutic developments in Friedreich Ataxia | Yvan Torrente (Milan)

12:10 Focusing on regulatory aspects in neuromuscular disease | Jan Smeitink (Nijmegen)

PARALLEL ROOM 1

10.30-12.30
POSTER SESSION PART II - NO CME

12:30 – 13:30
LIGHT LUNCH

PLENARY ROOM

13:30 – 14.30
SYMPOSIUM – SPONSORIZED BY ARGENX - NO CME

Exploring the role of efgartigimod from clinical trials to real practice in gMG
Chairpersons: Lorenzo Maggi (Milan) and Raffaele Iorio (Rome)

13:30 Welcoming

13:35 MG: an IGg mediated disease | Raffaele Iorio (Rome)

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

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

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

14:20 Discussion

14:30 – 15:30
SYMPOSIUM – SPONSORIZED BY ALEXION- NO CME

ECULIZUMAB: A YEAR OF VALIDATION FROM TRIAL DATA TO REAL WORLD EXPERIENCE
Chairpersons: Francesco Habetswallner (Napoli) and Michelangelo Maestri Tassoni (Pisa)

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

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

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

15:15 Discussion

15:30 – 16:00
COFFEE BREAK

16:00 – 17:30
Chairperson: TBD
PLENARY WORKSHOP 5
Idiopathic Inflammatory Myopathies: Diagnostic Challenges, New Entities, And Future Therapeutical Approaches

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

16:30 Immune checkpoint inhibitor–associated myositis: risk factor, disease courses and therapeutic approach | Antonello Farina (Rome)

17:00 Muscle MRI and imaging biomarkers: role on diagnosis and follow-up of IIM | Stefano Previtali (Milan)

17:30 – 18:30
SYMPOSIUM – SPONSORIZED BY AMICUS- NO CME

Come migliorare la Gestione della Malattia di Pompe
Charipersons: Tiziana Mongini (Turin) and Antonio Toscano (Messina)

17:30 Razionale di utilizzo delle small molecules nelle LSD | Giancarlo Parenti (Naples)
17:45 Presa in carico e follow up del paziente con malattia di Pompe | Cristina Sancricca (Rome)
18:00 Si può limitare il danno cellulare? | Antonio Toscano (Messina)

18:15 Discussion

18:30 – 19:30
General Assembly

Saturday 8 June 2024

PLENARY ROOM

08:30 – 09:30
Chairperson: TBD
Oral Communication | SMA

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

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

08:54 Fragility fractures prevalence and bone health in a large cohort of untreated SMA patients | Chiara Panicucci (Genua)

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

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

PARALLEL ROOM 1

08:30 – 09:30
Chairperson: TBD
Oral Communication | FSHD

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 (Rome)

08:42 Evaluation of non-invasive biological sources for assessing methylation levels of FSHD-associated locus | Claudia Strafella (Rome)

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

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

09:18 Molecular imaging of muscle involvement in facioscapulohumeral muscular dystrophy using multispectral optoacoustic tomography | Mauro Monforte (Rome)

PARALLEL ROOM 2

08:30 – 09:30
Chairperson: TBD
Oral Communication | Genomic

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

08:42 Recurrent TTN missense variants in biallelic titinopathies: a focus on Proline changes
Maria Francesca Di Feo (Genua)

08:54 Beyond exome-targeted sequencing approaches to increase the diagnostic rate of unsolved neuromuscular disease patients | Pasquale Di Letto (Naples)

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

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

PLENARY ROOM

09:30 – 10:30
Chairperson: TBD
Oral Communication | Mitochondrial

09:30 Developing AAV gene therapy for mitochondrial myopathies | Carlo Viscomi (Padua)

09:42Urolithin A as a potential treatment for mitochondrial myopthies | Valeria Balmaceda (Padua)

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

10:06 Serum neurofilament light chain in mitochondrial diseases: exploring a new promising biomarker. | Guido Primiano (Rome)

10:18 Role of muscle biopsy in mitochondrial myopathy: genotype-phenotype correlation
Dario Zoppi (Naples)

PARALLEL ROOM 1

09:30 – 10:30
Chairperson:TBD
Oral Communication | Duchenne

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

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

09:54 Gut microbiota signature in Duchenne Muscular Dystrophy | Chiara Panicucci (Genua)

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

10:18 Nonsense mutations in Becker muscular dystrophy: DMD gene position and disease severity | Domenico Gorgoglione (Padua)

PARALLEL ROOM 2

09:30 – 10:30
Chairperson: TBD
Oral Communication | Sperimentals

09:30 Creation of a Zebrafish model of LGMDD2 and role of TNPO3 in the pathogenetic mechanism Roberta Costa (Rome)

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

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

10:06 Modeling Myotonic Dystrophy type 2 in human cerebral organoids | Paola Spitalieri (Rome)

10:18 Patient-derived organoids as a new in vitro paradigm to study Myotonic Dystrophy type 1 Lorenzo Fontanelli (Pisa)

PARALLEL ROOM 2

10:30 – 11:30
Chairperson: TBD
Muscle TriviAIM – The Final

11:30 – 13:30
Chairperson: TBD
ROUND TABLE MEETING with PATIENT'S ASSOCIATIONS

13:30 Awards and conclusions

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