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12° CONGRESSO NAZIONALE ASSOCIAZIONE ITALIANA DI MIOLOGIA

LE MALATTIE MUSCOLARI: UN MODELLO PER LA DIAGNOSI, LA TERAPIA E LA PRESA IN CARICO DEI PAZIENTI CON MALATTIE RARE

17 – 19 Maggio 2012

Baia Samuele
Località Punta Sampieri - Scicli (Ragusa)

Giovedi', 17 maggio 2012

8.30 Registrazione dei partecipanti - Saluti delle Autorita'

9.00- 11.00 WORKSHOP - Miopatie dei Cingoli (LGMD): recenti acquisizioni cliniche e genetiche
Moderatori: G. Vita (Messina), M. Mora (Milano)

9.00-9.30 G.P. Comi (Milano) Eterogeneità clinica e genetica nelle forme autosomico recessive

9.30-10.00 C. Angelini (Padova) Forme dominanti: nuovi fenotipi e nuovi geni

10.00-10.30 G. Novelli (Roma) Aspetti clinici e genetici delle Laminopatie

10.30-11.00 G. D'Angelo (Bosisio Parini-LC) "Standard of care" e terapie sperimentali

11.00- 11.30 Coffee break

11.30-12.15 LETTURA MAGISTRALE - B. Schoser (Munich - Germany)
"Myotonic Dystrophy type 2- A phenotype summary of 300 families"
Moderatore: R. Massa (Roma)

12.15-13.00 COMUNICAZIONI ORALI : Canalopatie e Miotonie
Moderatori: G. Antonini (Roma), G. Meola (Milano)

12.15-12.30 **Clinical spectrum associated with mutations in *SCN4A* gene**
Maggi L. et al. (Milano, Brescia, Torino, Napoli, Como, Padova)

12.30-12.45 **High disease impact of skeletal muscle channelopathies on health status**
Sansone V. et al. (Milano, Londra, Torino, Messina)

12.45-13.00 **Clinical, genetic and muscle studies in myotonic dystrophy type 1 associated with variant CCG expansions.**
Santoro M. et al. (Roma)

13.00-14.00 Colazione di lavoro

14.00- 15.30 Visione e Discussione Posters
Moderatori: M. Mancuso (Pisa), S. Previtali (Milano), C. Terracciano (Roma),G. Di Jorio (Napoli), C.P. Trevisan (Padova), S. Corti (Milano), V. Sansone (Milano), E. Ricci (Roma)

15.30-17.0 Tavola rotonda con le Associazioni dei pazienti
"Centri di riferimento e presa in carico dei pazienti"
Moderatori: Ambrosini A (Milano), Politano L. (Napoli), T. Mongini (Torino)
Responsabili delle Associazioni:

M. Melazzini (AISLA)

F. Buccella, L. Genovese (Parent Project)

A. Fontana, A. Carbone (UILDM)

N. Riccobello (ASAMSI)

D. Lauro (Famiglie SMA)

R. Di Pietro (AIG)

P. Santantonio (Mitocon)

17.00- 17.30 Coffee break

17.30 - 18.45 COMUNICAZIONI ORALI: Miopatie metaboliche
Moderatori: S. Servidei (Roma), M. Filosto (Brescia)

17:30 – 17:45 **New quantitative MRI resources: Muscle Fat Fraction (MFF) examination of girdle and respiratory muscles and Its correlations with functional measures in a cohort of late onset GSDII patients.**
Barca E. et al. (Messina)

17:45 – 18:00 **Genetic characterization of a large cohort of McArdle patients**
Cassandrini D. et al. (Genova, Verona, Milano, Padova, Pisa, Messina, Roma, Monza, Torino, Brescia, Firenze, Napoli, Bologna, Conegliano-TV, Calambrone-PI)

18:00 – 18:15 **Genotype-phenotype correlation in Pompe Disease: a step forward**
De Filippi P. et al. (Pavia)

18:15 – 18:30 **New motor function's outcome measures during 1 year enzyme replacement therapy in 40 late onset GSDII patients**
Semplicini et al.(Padova, Pavia, Milan, Milan, Verona, Brescia, Roma, Milano, Palermo, Cagliari, Napoli, Torino, Messina)

18:30 – 18:45 **Symptomatic heterozygous patients in Late-Onset glycogen storage disease type 2**
Vercelli L. et al. (Torino, Pavia, Milano)

18.45-19.45 COMUNICAZIONI ORALI: Miopatie congenite
Moderatori: C. Minetti (Genova), M. Moggio (Milano)

18.45 – 19.00 **Clinical, morphological and genetic data in Italian patients with congenital myopathy**
Bruno C. et al. (Genova, Roma, Milano, Messina, Padova, Pavia, Torino, Napoli, Bologna, Calambrone-PI)

19.00 – 19.15 **Clinical, MRI, morphological and genetic characterization in centronuclear myopathy due to DNM2 mutations**
Catteruccia M. et al. (Roma, Milano, Napoli, Padova, Calambrone-PI, Genova)

19.15 – 19.30 **What prelamin A does to skeletal muscle?**
Mattioli E. et al. (Bologna)

19.30 – 19.45 **Whole exome sequencing as genetic diagnostic tool in Myofibrillar Myopathies**
Neri M. et al. (Ferrara, Roma, Verona, Rockville)

Venerdi', 18 maggio 2012

8.30 – 9.30 MUSCLE CLUB
Moderatori: C. Bruno (Genova), C. Rodolico (Messina)

8.30 – 8.40 **An unusual case of LGMD with an infantile onset**
Carboni N. et al. (Cagliari, Genova, Ferrara)

8.40 – 8.50 **Hemangioma of the semimembranosus muscle in a patient with late onset myopathy**
Cotelli M.S. et al. (Brescia)

8.50 – 9.00 **Myopathy or polymyositis?**
Manneschi L. et al. (Fidenza, Parma)

9.00 – 9.10 **A case of "Double trouble" in a myopathic patient**
Masciullo M. et al. (Roma)

9.10 – 9.20 **A massive vacuolar myopathy with an unusual phenotype and a strange molecular source**
Parisi D. et al. (Messina, Milano, Modena)

9.10 – 9.20 **Therapeutical challenges in a necrotizing myopathy**
Vercelli L. et al. (Torino)

9.30- 10.15 COMUNICAZIONI ORALI: Distrofie muscolari
Moderatori: G. Marrosu (Cagliari), M. Mirabella (Roma)

9.30 – 9.45 **Decorin deficiency in muscle of Ullrich Congenital Muscular Dystrophy and Myosclerosis Myopathy patients unrelated to collagen VI genes mutations**
Pellegrini C. et al (Ferrara, Bologna, Bagheria-PA)

9.45 – 10.00 **Scapular Girdle involvement in Facioscapulohumeral muscular dystrophy (FSHD): a MRI study**
Iannaccone E. et al (Roma)

10.00 – 10.15 **Collagen VI alpha 6 chain expression in skeletal muscle of Ullrich Congenital Muscular Dystrophy and Bethlem myopathy patient**
Tagliavini F. et al (Bologna, Ferrara, Bagheria-PA, Colonia)

INCONTRO TRA LE ASSOCIAZIONI SCIENTIFICHE AIM-SIAARTI
Moderatori: M. Ranieri (Torino), A. Toscano (Messina)

10.15-11.00 LETTURA MAGISTRALE - Z. Argov (Jerusalem - Israel) : "Practical approach to ICU acquired weakness"

11.00-11.30 Coffee break

11.30- 13.10 WORKSHOP - Problematiche anestesiologiche e rianimatorie nelle malattie muscolari

11.30-11.50 L. Morandi (Milano) Come valutare un paziente con iperCKemia

11.50-12.10 T. Mongini (Torino) Ipertermia Maligna e insufficienza d'organo nelle miopatie

12.10-12.40 F. Racca (Alessandria) Gestione peri- ed intra-operatoria dei pazienti con malattie muscolari

12.40-13.10 M. Antonelli (Roma) Approccio ventilatorio al paziente con malattia muscolare

13.15-14-15 Colazione di lavoro

14.15- 15.45 PROGETTI DI RICERCA E PROGRAMMI COLLABORATIVI
Moderatori: M. Moggio (Milano), R. Massa (Roma)

15.45-17.05 **WORKSHOP - "Next Generation Sequencing", tecnologie ed impatto nelle Malattie Neuromuscolari**
Moderatori: A. Ferlini (Ferrara), E. Pegoraro (Padova)

15.45- 16.10 R. Tanzi (Monza) - Tecnologia "Ion torrent": una rivoluzione nella rivoluzione.

16.10-16.35 L. Beretta (Milano) - Applicazioni cliniche della Next Generation Sequencing: ora , semplice, per tutti.

16.35-17.00 M. Neri (Ferrara) – " Whole-exome sequencing" per la diagnosi delle malattie muscolari

17.00-17.15 Coffee break

17.15- 18.00 COMUNICAZIONI ORALI : Atrofia Muscolare Spinale
Moderatori: L. Morandi (Milano), A. Berardinelli (Pavia)

17.15 – 17.30 **Targeted gene correction of Spinal Muscular Atrophy induced pluripotent stem cells and motoneurons as cell source for therapy**
Corti S. et al. (Milano)

17.30 – 17.45 **Phase-II multicenter double-blind placebo-controlled study of tolerability and efficacy of salbutamol in adult type III SMA patients**
Tiziano F.D. et al. (Roma, Milano, Padova, Torino, Messina, Napoli)

17.45 –18.00 **Oxidative defects at muscle histochemistry in 15 genetically-determined SMA cases**
Ripolone M. et al. (Milano, Medellin, Pavia)

18.00- 18.45 LETTURA MAGISTRALE - R. Tupler (Modena)
Distrofia muscolare Facio-Scapolo-Omerale: complessità genetica e nuove prospettive diagnostiche
Moderatore: C. Angelini (Padova)

18.45- 20.30 Assemblea dei soci

Sabato, 19 maggio 2012

8.30-9.30 COMUNICAZIONI ORALI: Distrofinopatie
Moderatori: A. Prelle (Milano), G. Nigro (Napoli)

8.30 – 8.45 **Search for SNPs modifiers in DMD with different corticosteroids response by candidate genes targeted resequencing**
Bovolenta M. et al. (Ferrara, Francoforte, Londra, Leiden, Newcastle, Roma, Rockville)

8.45 – 9.00 **Muscle fibrosis in the sgcb-null mouse model compared to the mdx model**
Gibertini S. et al. (Milano)

9.00 – 9.15 **Pilot study of flavocoxid in ambulant DMD patients**
Licata N. et al. (Messina)

9.15 – 9.30 **Germinal mosaicism and muscular dystrophies**
Tedeschi S. et al. (Milano, Napoli)

9.30-10.30 COMUNICAZIONI ORALI: Malattie Mitocondriali
Moderatori: G. Siciliano (Pisa), P. Tonin (Verona)

9.30 – 9.45 **MNGIE therapy: experience with continuous ambulatory peritoneal dialysis, allogeneic stem cell transplantation and carrier erythrocyte entrapped thymidine phosphorylase**
Filosto M. et al. (Brescia, Londra, Monza, Verona)

9.45 – 10.00 **Stimulation of the mitochondrial biogenesis is effective in mitochondrial myopathies**
Cerutti R. et al. (Milano)

10.00 –10.15 **Next-generation sequencing (NGS)-based molecular dissection of mitochondrial infantile hypertrophic cardiomyopathy and lactic acidosis (MIHCLA)**
Lamperti C. et al. (Milano)

10.15 – 10.30 MERRF - A genotype/phenotype study from the Italian Mitochondrial Registry
Siciliano G. and "The Italian Network of Mitochondrial Diseases"

10.30-11.15 LETTURA MAGISTRALE - S. DiMauro (New York – U.S.A.)
"Deficienza di CoQ10: fenotipi, genotipi e rimedi"
Moderatore: C. Messina (Messina)

11.15- 11.30 Coffee break

11.30- 13.10 WORKSHOP - Le alterazioni del SNC nelle malattie muscolari
Moderatori: E. Bertini (Roma), M. Zeviani (Milano)

11.30 – 11.45 **S. Messina (Messina) Alterazioni neuroradiologiche e cognitive dell'età evolutiva**

11.45-12.00 A. Berardinelli (Pavia) Le crisi epilettiche

12.00-12.15 V. Carelli (Bologna) Disturbi oculari

12.15-12.30 G. Meola (Milano) Alterazioni cognitive e correlati neuroradiologici nelle Miotonie Distrofiche e non-Distrofiche

12.30-12.45 O. Musumeci (Messina) Esistono i disturbi extrapiramidali nelle malattie muscolari ?

13.10 Conclusione del congresso

SESSIONI POSTER: Canalopatie e Miotonie

P1. Sleep breathing disorders in Myotonic Dystrophy type 1 (DM1) and 2 (DM2)
Bianchi M.L.E. et al. (Roma)

P2. A Roman Network for the Myotonic Dystrophies: start-up and construction of a patients' database
Bucci E. et al. (Roma)

P3. Vitamin D deficiency and falls in myotonic dystrophies
Bugiardini E. et al. (Milano)

P4. Respiratory status at diagnosis in Myotonic Dystrophy type 1
Cagnetti C. et al. (Ancona)

P5. Correlation of ribonuclear inclusions size with biomolecular and clinical findings in myotonic dystrophy type 2
Cardani R. et al. (Milano, Pavia, Roma)

P6. Myotonia congenita with an atypical phenotype and a novel CLCN1 gene mutation
Portaro S. et al. (Messina)

P7. Correlation between CUGBP1 expression to biomolecular, clinical and histopatological phenotype in DM1 and DM2 patients
Renna L.V. et al. (Milano, Roma)

P8. The frequency and severity of cardiac abnormalities are reduced in myotonic dystrophies type 2 (DM2) compared to type 1
Sansone V. et al. (Milano)

P9. Vitamin D deficiency in myotonic dystrophy type 1
Terracciano C. et al. (Roma)

SESSIONI POSTER: Malattie Mitocondriali

P10. New genotype-phenotype correlations in subjects with POLG1 mutations
Borgione E. et al. (Troina-EN)

P11. Unusual phenotype associated with the 3271T>C mutation in the mitochondrial tRNALeu(UUR) gene
Brisca G. et al. (Genova, Calambrone-PI)

P12. SANDO/MNGIE-Like overlap syndrome due to polymerase gamma mutation
Buono R. et al. (Verona, Udine, Brescia)

P13. Successful intravenous immunoglobulin treatment in a mitochondrial patient with acute respiratory failure
Caldarazzo Ienco E. et al. (Pisa)

P14. Sporadic myopathy, myoclonus, leukoencephalopathy, neurosensory deafness, hypertrophic cardiomyopathy and insulin resistance associated with the mitochondrial 8306 T>C MTTK $tRNA^{Lys}$ mutation
Cardaioli E. et al. (Siena, Parigi)

P15. T10158C mutation in ND3 gene causes mitochondrial overlap syndrome: a case report
Carluccio M. A. et al. (Siena, Perugia, Spoleto)

P16. Markedly effective gene therapy in an Ethylmalonic Encephalopathy mouse model
Di Meo I. et al. (Milano, Napoli, Padova)

P17. Mutations of the mitochondrial-tRNA modifier MTO1 cause hypertrophic cardiomyopathy and lactic acidosis
Ghezzi D. et al. (Milano, Parma, Neuherberg, Monza, Padova)

P18. Microscaleoxygraphy unveils respiratory chain defects in cells from mitochondrial disease patients
Invernizzi F. et al. (Milano, Copenhagen, Pavia)

P19. MELAS-like encephalomyopathy caused by a new pathogenic mutation in mitochondrial DNA encoded cytochrome c oxidase subunit I
Lamperti C. et al. (Milano)

P20. Association between ischemic lactate and NADH:ferricyanide-reductase muscle activity
Orsucci D. et al. (Pisa)

P21. Centronuclear myopathy with ragged red fibers and mtDNA multiple deletions: a case report
Ricci G. et al. (Pisa, Milano, Roma)

P22. Sengers syndrome: clinical, morphological and genetic features in two families
Robbiano A. et al. (Genova, Calambrone-PI)

P23. A novel mithocondrialtRNA^{Hys}point mutation in a patient with PSP-like phenotype
Romeo S. et al. (Messina)

P24. Hyperckemia as isolated feature of mitochondrial G5540A *tRNA^{Trp}* gene mutation
Russignan A. et al. (Verona, Brescia)

P25. THE ROLE OF BRAIN MRI IN MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOMYOPATHY
Scarpelli M. et al. (Verona, Brescia)

SESSIONI POSTER: Miopatie Metaboliche

P26. An unusual clinical presentation of Pompe disease characterized by a severe distal myopathy
Biasini F. et al. (Messina)

P27. Geno-phenotyping of a large Italian family with late-onset glycogenosis II
Cipullo F. et al. (Napoli, Messina, Padova)

P28. A family with epilepsy, choreo-athetoid movements, mental retardation and exercise-induced rhabdomyolysis: expanding the spectrum of GLUT-1 Deficiency Syndrome ?
Montagnese F. et al. (Messina, Catanzaro, Reggio Calabria, Cosenza)

P29. Trunk muscle involvement in 21 patients with late-onset Pompe disease
Ravaglia S. et al. (Pavia)

P30. A novel mutation in CPT2 gene in two unrelated patients from Campania region
Taglia A. et al. (Napoli, Milano)

P31. Bone pathology in Pompe disease: high incidence of micro-fractures in a group of patients without risk factors for osteoporosis
Zappini F. et al. (Verona, Brescia, Padova)

SESSIONI POSTER: Atrofie Muscolari Spinali

P32. Proposal of a structured protocol for the assessment of bone metabolism and bone mineral density in children with SMA
Baranello G. et al. (Milano, Pavia)

P33. Bone health determinants in spinal muscular atrophy
Brigati G. et al. (Genova)

P34. Genotype- phenotype correlation in dHMN with infantile onset.
Fiorillo G. et al. (Calambrone-PI, Bologna, Milano, Pavia, Genova)

P35. Sleep-wake cycle in the Kennedy's disease
Liguori C. et al. (Roma)

P36. Mechanical cough assist in SMA1 patients under 1 years of age
Pedemonte M. et al. (Genova, Milano)

P37. Normal cardiac function in Kennedy's disease
Querin G. et al. (Padova, Milano, Novara, Verona)

SESSIONI POSTER: Distrofinopatie e altre distrofie muscolari

P38. Earlier cardiomyopathy onset in DMD patients carrying the TT genotype AT *SPP1* rs28357094
Barp A. et al. (Padova, Milano, Napoli, Messina, Washington DC)

P39. A novel PTRF-CAVIN mutation in a patient with congenital lipodystrophy and mild muscular dystrophy
Bragato C. et al. (Milano, Bergamo)

P40. Whole Exome Sequencing and RNAseq in a Duchenne-like female with no dystrophin mutations: search for dystrophin gene modifiers
Brioschi S. et al. (Ferrara, Roma, Bari, Rockville)

P41. Role of calcineurin/NFAT on muscle regeneration in dystrophic process
Cama A. et al. (Messina)

P42. Pitfalls in FSHD molecular diagnosis: FSHD2 and undetected D4Z4 repeats contractions
Cao M. et al. (Padova, Nizza, Leiden)

P43. A clinical entity with muscular dystrophy, partial lipodystrophy, hypertriglicemia and diabetes in search for the causative gene
Carboni N. et al. (Cagliari, Roma, Monserato)

P44. Non progressive dysferlinopathy: benign course or response to steroids?
Cuccagna C. et al. (Roma)

P45. Implication of SIRT1 and its downstream pathways in dystrophic process
De Pasquale M.G. et al. (Messina)

P46. Implication of SIRT1 and its downstream pathways in dystrophic process
Falzarano M.S. et al. (Ferrara, Bologna, Alessandria, Padova, Leiden)

P47. Vitamin D and Bone Mineral Status in children affected by Duchenne Muscle Disease (DMD) treated with glucocorticoid: preliminary data
Inguaggiato E. et al. (Calambrone-PI)

P48. Body composition and muscle strength of individuals with Bethlem myopathy and Ullrich congenital muscular dystrophy
Miscione M.T. et al. (Bologna)

P49. Deletion of TRIM 32 gene in compound heterozygosis with a stop mutation: report of a case
Neri M. et al. (Ferrara, Verona)

P50. lower limb muscle involvement as clinical presentation of FSHD: a clinical study on 122 CASES
Pastorello E. et al. (Padova)

P51. Estimating body composition in adult individuals with Bethlem myopathy: comparison of bioelectrical impedance analysis and skin fold-thickness measurement
Pellegrini M. et al. (Modena, Bologna)

P52. Body composition and resting energy expenditure of adult individuals with Bethlem myopathy and Ullrich congenital muscular dystrophy
Pellegrini M. et al. (Modena, Bologna)

P53. Intragenerational phenotype-genotype variability in FSHD: the D4Z4short fragment might not be enough
Peverelli L. et al. (Milano, Piancavallo, Helsinki, Modena)

P54. A novel homozygous beta-sarcoglycan gene mutation: case description
Ricci G. et al. (Pisa, Napoli)

P55. Slightly defective α-dystroglycan glycosylation in a patient with mild limb girdle muscle dystrophy due to novel POMT2 mutations
Saredi S. et al. (Milano)

P56. Congenital muscular dystrophy with *FKRP* mutation
Trovato R. et al. (Calambrone-PI)

P57. Effects of steroids on Left Ventricular Ejection Fraction in cardiopathic DMD patients
Trucco F. et al. (Genova)

P58. X-Chromosome inactivation pattern in Duchenne muscular dystrophy carriers
Viggiano E. et al. (Napoli)

P59. DMD fibroblast nodules: an *in vitro* model potentially useful for high-throughput screening in muscle fibrosis
Zanotti S. et al. (Milano)

SESSIONI POSTER: Miastenia, Miopatie Congenite, Miositi, Varie

P60. Inflammatory myopathy and systemic sarcoidosis in a patient with myasthenia gravis
Alfonzo A. et al. (Messina)

P61. Congenital myopathy associated to MHY2 mutation: Italian case report
D'Amico A. et al. (Roma)

P62. A study on ventilatory device effectiveness and impact on patients quality of life: ARIA project
Fossati F. et al. (Milano)

P63. "AT SCHOOL WITH NEMO" an educational and skill training project for caregivers of children affected by Neuromuscular Disorders (NMD's).
Gorni K. et al. (Milano)

P64. Adiponectin induces proliferation and enhances skeletal muscle differentiation of IBM mesoangioblasts in vitro and in vivo
Lucchini M. et al. (Roma)

P65. A novel intronic LaminA/C substitution associated with a muscle pattern suggestive for myofibrillar myopathy
Magri F. et al. (Milano)

P66. Whole Exome Sequencing filtered by novel candidate genes as tool for gene discovery in a recessive family with Parkinson and ataxia
Neri M. et al. (Ferrara, Verona, Roma)

P67. Modeling Alpha-Dystroglycanopathies in Zebrafish
Pappalardo A. et al. (Calambrone-PI, Firenze, Genova)

P68. Clinical and pathological work up in a case of infantile "idiopathic" toe walker.
Pini A. et al. (Bologna)

P.69. A 80-year-old man with myalgia, hyper-CK-aemia, hypocoagulability and vitamin-D deficiency
Pizzolato R. et al. (Roma)

P70. Life-threatening dysphagia as the only manifestation of sporadic inclusion body myositis (sIBM)
Primiano G. et al. (Roma)

P71. The mandatory role of muscle biopsy in sporadic IBM: a case report
Salaroli R. et al. (Bologna)

P72. Congenital myastenic syndromes due to a novel GFPT1 mutation: a new Italian case
Sframeli M. et al. (Messina, Monaco, Newcastle)

P73. Amyloid myopathy mimicking polymyositis
Velardo D. et al. (Milano)