I-1
History of Myology in Italy and its international collections
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The history of Myology in Italy begins in 16th century with muscle anatomy pictures by Vesalius and Canani while in the 19th century the Neapolitan physician Gaetano Conte described Duchenne muscular dystrophy on clinical basis (1).

Myology reached a major development in the 20th century with the establishment of the CNR center in Padova (professor Massimiliano Aloisi) a cardiomyological and genetic center in Naples (professor Giovanni Nigro) and a neurological center in Milan (professor Guglielmo Scarlato). In the 60th prof. Aloisi and Federico Milcovich, a myodystrophic patient, started the Italian Muscular Dystrophy patients association (UILDM) with contacts with MDA (2). The first congress in Neuromuscular diseases took place in Milan in 1969 by the organization of professor Scarlato, Aloisi, Canal and was attended by several outstanding international muscle researchers such as AG Engel, WK Engel, LP Rowland, M.Fardeau and I.Hausmanowa-Petrusewicz. Eleven meetings in many countries followed this, up to the last XII international Congress on Neuromuscular Diseases in Naples in 2010.

Several laboratories arose in the country especially in Neuromuscular Institutes; several researchers emigrated permanently or went for a stage to improve their myological skills especially in USA, UK, Canada, France both in basic and/or pathological, clinical research (Table 1). The laboratory of Columbia in New York, lead by professor Di Mauro was a common place of training especially in the field of mitochondrial myopathies.

As in other developed countries, the basis of neuromuscular research and diagnosis were expanded on immunohistochemical, biochemical and molecular grounds, during the 20th and the beginning of the 21st century. Enzo Ferrari – a race car factory engineer – was a support to research in muscular dystrophy in Milan, Padova and Modena. Telethon has contributed to support in neuromuscular disorders. Myology with the enlargement of scientific basis in the molecular era began to split in sub-specialities i.e. genetics, physiopathology etc. and a spectrum of knowledge was accumulated both for diagnostic and therapeutic purposes. In the field of metabolic diseases and limb-girdle myopathies several Italian laboratories described new entities. A number of treatments, beside the treatment of inflammatory myopathies and myasthenia gravis, were found and applied in metabolic myopathies: i.e. carnitine, Coenzyme Q, enzyme replacement in glycosogenosis type II.

The continuous challenge of the treatment of the primary muscular dystrophy remains for the future since so far there are only emerging molecular therapies: antisense oligonucleotides in DMD, adeno viral therapy in sarcoglycanopathies, cell therapy might contribute to answer to a promises for muscular patients.

In the translation area many laboratories and Italian groups continue with new researchers and they have contributed to meetings of European Neuromuscular Center and to the Foundation and organisation of the World Muscle Society with meetings in Italy (Naples, Taormina), and to the Treat – NMD and Eurobiobank networks.

References
1. Angelini C. Handbook of clinical neurology - History of Neurology. In: Finger S, Boller F, Tyler KL, eds. Muscular Dystrophy. Vol. 95. Amsterdam: Elsevier 2010, pp. 477-88.
2. Angelini C, Aloisi M. Obituary. Neuromusc Disord 2000:10:1-2.

I-2
Can we bypass a muscle metabolic defect?
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The metabolic myopathies are thought to be more amenable to treatment once the defect in the biochemical pass way is identified. Some of the defects cause mainly exercise intolerance and its improvement can be regarded as therapeutic success. In others muscle weakness and degeneration is seen and functional and strength improvement is the goal.

Therapy of such muscle metabolic disorders can be achieved through enzyme replacement, e.g. therapy of acid maltase deficiency (Pompe’s disease) which is currently administered to young and adult patients. Enzyme upregulation can also be attempted and a case in point is bezafibrate in carnitine palmityl transferase 2 deficiency.

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Table 1. The main international connections.

| Country | Laboratory/Institute | City | Founders/Researchers |
|---------|---------------------|------|---------------------|
| USA     | Mayo Clinic (Engel AG) | Chicago | Angelini, Mora, Milone, Fumagalli |
| USA     | USC Los Angeles (Engel WK, Askanas) | Los Angeles | Martinuzzi, Vita, Broccollini, Mirabella, Valterini |
| USA     | Columbia University (Rowland, Di Mauro) | New York | Trevisan, Bresolin, Bruno, Mancuso, Zeviani, Servidei, Ricci, Minetti, Moggio, Musumeci, Salvati |
| Canada  | Montreal (Karpatsky) | Montreal | Armani |
| UK      | London (Dubowitz) | London | Muntoni, Mercuri, Sorarù |
| UK      | London (Moran-Hughes) | London | Toscano |
| UK      | Newcastle (Walton, Bushby) | Newcastle | Vila, Gugliere |
| UK      | Oxford (Vincent) | Oxford | Evoli |
| UK      | Liverpool (Edwards) | Liverpool | Siciliano |
| France  | Paris (Fardeau, Tomè) | Paris | Villanova, Berardinelli |
| France  | Nice (Desnuelle) | Nice | Sacconi |
| France  | Poitiers (Rideau) | Poitiers | Nigro, Comi, Politano, Bianchi |
| Poland  | Warsaw (Hausmanowa-Petrusewicz) | Warsaw | Nigro, Comi, Petrella, Politano |