EDITORIAL

The MDR '95 Conference

Fifteen years ago we organized in Venice, with Proff. Danieli and Fontanari, the first International Scientific Meeting dealing with the advances and new trends in Muscular Dystrophy Research: MDR '80. The Proceedings (Elsevier International Congress Series 527) contained the description of Duchenne dystrophy, Becker muscular dystrophy and of severe autosomal limb girdle muscle disorder that was found in Tunisia. Two subsequent meetings, MDR '90 and MDR '93, were held at the International School of Neurological Sciences in S. Servolo, and focused on the molecular diagnosis of muscular dystrophies (Excerpta Medica, International Congress Series 934; Neuromuscular Disorders 1994).

In the last ten years, an unprecedent advance took place in understanding muscle disorders. We know now that Duchenne and Becker muscular dystrophy are due to either absence or defective dystrophin (Table 1) and severe autosomal recessive muscular dystrophies are due to the absence of dystrophin associated glycoproteins (DAG) which are components of the sarcoglycan complex (adhalin or 50 kDa DAG named α-sarcoglycan, β-sarcoglycan or A3b, γ-sarcoglycan or 35 kDa DAG).

Therefore, defects of DAG's result in various types of muscular dystrophy: for example the Tunisian autosomal limb girdle dystrophy is due to γ-sarcoglycan mutation; there are also primary and secondary adhalin defects. A new mutation in adhalin gene is presented in this issue that results not only in myopathy but also in cardiopathy. The basal lamina is connected with dystrophin and is composed of a complex network, including collagen fibers, laminin and heparan sulphate proteoglycan (Fig. 1). It has been shown in the last years that a defect in merosin (M-laminin) can lead to congenital muscular dystrophy (CMD). This information allows to detect a precise pathogenesis and prenatal diagnosis in the western form of CMD. Western patients often have central nervous system involvement, since merosin is also expressed in Schwann cells. Treatment of myopathies seems closer now that pathogenesis is better understood. The unravelling of complexity of sarcoglycan and dystroglycan complex is sheding light on numerous muscle dystrophies and may lead to new therapeutical strategies.

Our group observed that boys with Duchenne dystrophy develop better muscle strength and have a longer period of walking when treated with Deflazacort (a new synthetic steroid), with less side effect than Prednisone. Future pathogenetic developments are needed to improve the results of steroid treatment and minimize side effects. Some new gene transfer techniques have been presented at the meeting with the biolistic approach in mdx mice. Further developments in gene transfer vectors let us hope that muscle research may soon bring some relief to patients suffering from muscle disorders, still waiting for therapy. Among the treatable neuromuscular disorders, such as polymyositis and myasthenia gravis, new exciting therapeutic advances were presented at MDR '95. An up-to-date in the field of myasthenia gravis was the presentation of a

Table 1. Muscular dystrophies: clinical presentation and recognised defects.

CLINICAL PHENOTYPE	MOLECULAR DEFECT
1. DUCHENNE MUSCULAR DYSTROPHY	DYSTROPHIN DEFECT
2. BECKER MUSCULAR DYSTROPHY AND MILDER PHENOTYPES: MYALGIA, CRAMPS CARDIOMYOPATHY, MYOGLOBINURIA	DYSTROPHIN ABNORMALITY
3. SEVERE AUTOSOMAL RECESSIVE MUSCULAR DYSTROPHY (SCARMD)	α - SARCOGLYCAN (ADHALIN) DEFECT
4. EARLY ONSET DYSTROPHY	β - SARCOGLYCAN (A3b) DEFECT
5. EARLY LIMB-GIRDLE MUSCULAR DYSTROPHY	γ -SARCOGLYCAN DEFECT
6. CONGENITAL MUSCULAR DYSTROPHY WESTERN-TYPE	MEROSIN DEFECT

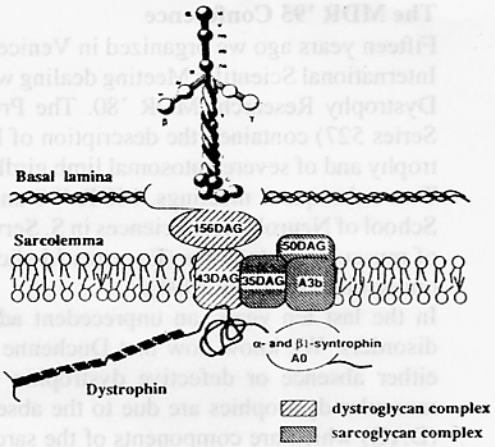

Figure 1. Dystrophin, sarcoglycan and dystroglycan complex and its interaction with merosin (figure modified from Sugita H., Am J Pathol, 1994).

new endoscopic technique of thymectomy and in severe steroid-resistant myasthenia gravis, the use of cyclosporine A. Polymyositis and acute or chronic demyelinating neuropathy profit from various immunosuppressant regiments and of IVIg (high dose intravenous immunoglobulines). The most severe and devasting neuromuscular disorder, i.e. amyotrophic lateral sclerosis, is undergoing new therapeutic trials. The results with recombinant insulin-like growth factor (FGF-I) in motor neuron disease were presented. The potential use of growth factors in treatment of neuromuscular diseases has to be evaluated in other muscle disorders. New molecular and therapeutic approaches of glycogenosis type V and lipid storage myopathy were presented, in which metabolic replacement with carnitine and riboflavin represent an effective therapy and may be added to the prescribed diet.

The present issue of BAM contains the abstracts and selected papers of the Meeting MDR '95 held in Padova University in November 1995, sponsored by the Regional Neuromuscular Center, Sigma-Tau and UILDM (Unione Italiana Lotta alla Distrofia Muscolare). The papers collected in this issue of BAM confirm that at the University of Padova there is a clinical muscle group with several collaborators in other Italian Universities and international Centers. The most encouraging results are the progress in diagnosis and therapy in many muscle diseases that were obtained by clinical research. The active discussion and several contributions depicted an impressive outlook of very productive research in this field.

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