I-15
Neuromyotonia: Potassium channelopathy or non ionic disease?

G. Serratrice
CHU La Timone, Marseille, France

The early pathophysiologic study showed increasing evidence that autoimmunity is implicated in the pathogenesis of neuromyotonia. Antibodies to voltage gated potassium channel were detected in the serum of patients who had peripherical nerves hyperexcitability and also Morvan's disease or limbic encephalitis. These discoveries offered new approaches to treatments.

Recently, antibodies previously attributed to VGKC recognise 2 surface antigens LGI1 and CASPR2 into the VGKC complex. Finally, VGKC antibodies are directed to 2 proteins the first one is a key hippocampic protein containing pre and post synaptic proteins. The second one CASPR2 is an hippocampic and paranodal protein. There clinical significance is different: hyperexcitability limbic encephalitis without thymoma for LGI1, hyperexcitability Morvan limbic encephalitis and frequent thymoma for CASPR2.

In conclusion, the term NMT - LE - VGKC should be changed to NMT- LE with LGI1 and CASPR2 antibodies and classified as auto immune synaptic disorders.

I-16
Crossing barriers: how medical doctors and researchers can partner with Advocacy Groups

L. Feld
Periodic Paralysis Association Vice President, Orlando, Florida, USA

Disease advocacy groups (DAGs) by and large arise in chronic or devastating diseases for which persistent unmet needs exist. Patients seek DAGs for a variety of reasons, but most importantly to find immediate psychological, diagnostic, and therapeutic coping mechanisms. Factors that drive their continued involvement include hope for finding a permanent cure and an altruistic desire to increase awareness and share their experiences to help others in similar need. In doing so, they frequently establish a conduit to world experts in their rare disorder, and where needed, better define disease characteristics and management.

DAGs provide help to their members in a variety of ways:

1) offer web-based, easily accessible and medically reliable information to patients about their condition;
2) offer a means of members to interact either through email list serves, blogs, or message boards;
3) organize annual meetings for members, whereby disease experts and members can interact face to face;
4) offer a virtual interface between medical experts and their membership via on-line "Ask The Experts";
5) potentiate blood banking and genetic research by gathering relatively large numbers of patients in one repository;
6) organize and galvanize scientific and medical experts to research the condition;
7) motivate experts to help the DAG members.

Physicians are the ultimate backbone of any DAG that wishes to provide more than psychosocial coping modalities for their members. Members ultimately seek credible medical knowledge as the best chance to navigate their illness and achieve normalcy in their lives. The fundamental ways in which physicians can support a DAG is by being actively available to attend DAG member conferences, answer member questions, contribute to website content, and cooperate to establish consensus protocols for diagnosis and therapy, and work with each other to find meaningful diagnostic modalities and treatments.

Barriers to a DAG's achieving its mission are many. DAGs often lack funding, frequently suffer from disinterest by a majority of the few medical experts interested in their disease, are sometimes hurt by splinter groups for the same disease fragmenting limited resources, and may find themselves caught between political or personality differences among their medical experts.

Barriers to physician participation in DAGs are many: competition for time of the physician by other diseases, limited time to deal with individual patient concerns, medico-legal uncertainties of interactions with DAG members, lack of recognition or peer reviewed publication credit for writing website content, cost of travel to meetings, having only non-clinical research interest, and knowledge in only a limited aspect of a disease in question.

I-17
Functional evaluation of muscle impairment in neuromuscular disorders

E. Mazzone
Not arrived

I-18
Chronic muscle electrical stimulation in Myotonic Dystrophy

C. Chisari
Not arrived

I-19
Neuromuscular electrical stimulation training in facio-scapulo-humeral muscular dystrophy

S. Sacconi
Not arrived

I-20
Wearable monitoring systems and rehabilitation

D. De Rossi
Not arrived

I-21
Current therapeutic guidelines in Duchenne Muscular Dystrophy to prolong life

V. Riccio, I. Riccio, F. Gimigliano, D. Riccardi, G. Riccardi, R. Gimigliano, L. Politano[1], G. Nigro[1], G. Duport[1], Y. Rideau[1]
Dipartimento di Scienze Ortopediche, Traumatologiche, Riabilitative e Plastico Ricostruttive Second University of Naples; [1] Institut Duchenne Italia (IDI), Naples, Italy

Duchenne's myopathy is an X-linked disease with well defined evolutionary phases, characterized by degradation of the

walking function, development of evolutive scoliosis and progressive decline of the respiratory function leading patients to premature death.

In 1985 Y. Rideau in France carried out a new global therapeutic strategy for treatment of lower limb deformities, scoliosis deformity and progressive restrictive syndrome.

The indication for surgery at the lower limbs is made very early, at the onset of the first signs of disease. The procedures are carried out at the same time and always bilaterally; they include: (i) hip section of superficial flexors; (ii) iliotibial band resection; (iii) subcutaneous tenotomy of semitendineous and gracilis; (iv) subcutaneous lengthening of Achilles tendons.

In the post-operative period, the patient begins exercises of active and passive mobility in few days and after three weeks recovers his performances; ambulation will remain almost normal for several years. A comparison of two groups of patients, the first precociously operated on the lower limbs, the other one not operated, shows better performances in the operated group.

The indications for surgical treatment of Duchenne scoliosis must be made after the loss of ambulation and not too late, to avoid the concurrent respiratory restrictive syndrome makes the patient inoperable. Over ten years ago, in Poitiers, a specific instrumentation for Duchenne scoliosis was created, providing for cylindrical rods fixed by peduncular screws at the sacro-lumbar level. On the dorso-lumbar level, the rod becomes flat to allow more flexibility of the trunk. The complications observed in a group of 55 patients operated for scoliosis, consisted in 2 cases of breaking of rods and 1 superficial infection. The surgery approach in DMD has the double aim to prolong the time of the autonomous ambulation and to avoid the evolution of scoliosis, limiting the harmful effects of the scoliosis on the respiratory function.

However, the surgery alone is unable to prolong the life expectancy in these patients, without treating the restrictive respiratory syndrome, first by nasal ventilation and then by elective tracheotomy, essential for the survival of the patient.

I-22
Early indicators of respiratory impairment in muscular dystrophies: from bioengineering to rehabilitation

M.G. D'Angelo

Not arrived

I-23
Respiratory rehabilitation in Duchenne Muscular Dystrophy

G. Fiorentino, A. Annunziata, R. Cauteruccio, M. Caputi
Cardiorespiratory Department, Monaldi Hospital, Second University of Naples, Italy

Lung function in Duchenne Muscular Dystrophy (DMD) is complicated by a decline in vital capacity (VC). After an initial period of normal increase in VC, that typically reach a plateau between 12 and 16 years, there is a period of decline, estimated in 200ml/year. The deterioration of the respiratory function (restrictive syndrome) is one of the two major causes of death in these patients and a close correlation between the development of respiratory symptoms and VC has been observed. An indirect consequence of intercostals muscles (IM) weakness is a reduction in respiratory system compliance (RSC), the ratio of the change inspiratory system volume to a change in pressure (RSC = AV/AP). Mobilisation of secretions, by postural drainage, is an important step in the clinical pathway of airway clearance and of vital importance in DMD. Therapy should be provided for no longer than necessary to obtain the desired therapeutic results. The use of airway clearance techniques, including assisted coughing techniques, both manual and mechanical, is strongly recommended. These techniques should always be included in the treatment of chronic NMD patients. Cough can be assisted by manual and mechanical means. All methods require a combination of improved insufflation of the lungs to achieve sufficient lung volumes for an effective cough in conjunction with adequate forced expiratory techniques to increase the patient's natural, but weakened, cough. Glossopharyngeal breathing (GPB) is the act of the glottis taking air and propelling it into the lungs. Some authors affirm that GPB can sustain normal ventilation throughout daytime hours without using a ventilator, and safely in the event of ventilator failure during sleep in patients with reduced inspiratory muscle function. In our opinion there is not enough evidence for generalised use of GPB alone to avoid invasive ventilatory assistance.

The cough assist machine (the mechanical insufflator-exsufflator) has proven to be useful to airway clearance in patients with neuromuscular weakness. It provides both an inspiratory phase (to inflate the lungs) and an expiratory phase (for the actual cough), all in one piece of equipment. Studies have shown that the Cough-Assist machine is well tolerated, without increased risk for complications such as pneumo-thorax, gastro-esophageal reflux, or pulmonary hemorrhage. It also can be used through various interfaces such as mouth, full face mask, or endotracheal or tracheostomy tube.

The indications for tracheostomy include: 1) NIPPV no longer effective at treating hypoventilation; 2) NIPPV not tolerated by the patient; 3) Excessive oral secretions; 4) Resources for outpatient management with NIPPV not available in the community; 5) Failure to extubate patient who has been intubated for whatever reason. In our experience tracheostomy is performed in election, when the vital capacity is about 700-1000 ml, so that the patient can long remain without constant ventilatory support.