

XIth International Congress on Neuromuscular Diseases

Istanbul, Turkey, 2-7 July, 2006.

Among four teaching courses, 25 plenary lectures, 14 focused symposiums and 10 workshops, over 150 oral presentations, more than 300 posters, plus two drug company sponsored satellite symposia, spread over six days and four floors of the grand Lutfi Kirdar Convention and Exhibition Centre in Istanbul, I can only present selected highlights, which may be biased towards personal interest, and, at least in part, my inability to be in more than one place at one time.

Muscle

The early morning plenary lectures ranged from basic sciences/biology to clinical and research in myopathies/muscular dystrophies. A special session was devoted to Dr Duchenne de Boulogne's 200th centenary and was given by the eminent French neuropathologist, Michel Fardeau. The work of Duchenne was really impressive by its scope, as well as by the number of technical innovations that made it possible: electrical stimulation of the muscles, muscle biopsy samples studied microscopically and finally, photography for the analysis of the physiology of facial expression. His achievements are still more impressive when one considers that they were those of a simple medical practitioner, whose life had very difficult moments.

Novel ways to treat muscular dystrophy (especially Duchenne type) may include axon skipping, stop codon read-through, upregulation of utrophin, myostatin inhibition, myogenic cell transplantation. However, significant technical challenges remain regarding their use in humans and these approaches will take several more years before being widely available in a clinical setting.

Neuromuscular junction

Investigation of congenital myasthenic syndromes (CMS) has disclosed a diverse array of molecular targets. Andrew Engel linked the clinical, electrophysiologic, and morphologic studies of endplates for detecting CMS-related



mutations in each subunit of presynaptic, synaptic, postsynaptic receptor defects, including MuSK as a new target for mutations which decrease the density of AChR on the junctional folds. John Newsom-Davis delivered a succinct overview of MG and pointed out there are still many unresolved issues such as whether thymectomy is beneficial in the non-thymomatous MG patient. This debate may be resolved by an ongoing large multicentre international single blind randomised clinical trial (RCT) of thymectomy. RCTs of mycophenolate mofetil in generalised MG and prednisone treatment in ocular MG are also ongoing.

Bertrand Fontaine delivered an excellent review of exercise tests coupled to electromyography to increase the sensitivity of the diagnostic procedure for channelopathies, enabling prediction for groups of mutations that can be subsequently directed by molecular diagnosis.

Peripheral nerve

P James Dyck Jr from the Mayo clinic gave an account of the utility of targeted proximal fascicular nerve biopsies from abnormal sites demonstrated by MRI (3 Tesla). Their experience showed proximal biopsy, despite being invasive, may be justifiable because of low morbidity and significant therapeutic implications.

Although some progress has been made in the evidence base for treatment of uncommon

peripheral nerve disorders, most notably the inflammatory neuropathies, no trials with neuroprotective and neuroregenerative agents have been concluded yet. Experimental trials have shown, however, that erythropoietin prevents distal axonal degeneration in a mouse model of taxol-induced neuropathy (John Hopkins group).

Amyotrophic lateral sclerosis(ALS) / Spinal muscular atrophy (SMA)

Since its description by Charcot, the mechanism of selective death of motor neurons in ALS has remained elusive. DW Cleveland from San Diego reported the use of antisense oligonucleotides to interfere with expression of the abnormal SOD1 gene in models of familial ALS. The authors are now preparing to test this treatment in primates and the first (phase 1) clinical trial may be ready to start in 2007.

Whereas SMA is an inherited neuromuscular disorder which affects all components of motor unit, the gene SMN, which codes for SMN protein, is either missing or reduced. Kathryn Swoboda (Utah) presented data from two completed open label studies of several compounds which enhance SMN expression in patient cell lines or prolong survival in an SMA animal model.

Conclusion

Some of the advances presented at this congress had immediate clinical relevance. The best management of the neuropathic consequences of glucose intolerance or metabolic syndrome is by treating the underlying cause. Also, prednisolone usage in neuralgic amyotrophy and enzyme replacement therapy in Fabry's neuropathy were presented. In the future, conditions such as ALS or DMD may have mechanism-specific disease-modifying treatments, as they share the final common pathway of endoplasmic reticulum stress induced proteins.

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CONFERENCE PREVIEW: Epilepsy Specialist Nurse Association (ESNA)

Birmingham, UK, 22-24 November, 2006.

The 2006 ESNA annual conference offers a two and a half day programme which provides support, information and advice to health care professionals caring for people with epilepsy. It will include workshops by epilepsy nurse specialist in paediatrics Pipa Hall, who will demonstrate a project that increased epilepsy awareness in the education system through theatre/acting. Phil Tittensor, epilepsy nurse specialist in adults, will present the research he has undertaken in complementary therapies in epilepsy. Dr Simon Nightingale, a consultant neurologist, will discuss his experience in taking a neurological his-

tory. Dr Hugh Richards, a neuropsychiatrist, will discuss mental health issues and epilepsy. Alice Hanscombe will lead on how to develop skills on telephone work.

Platform presentations include Beth Irwin, epilepsy nurse and midwife, who will update us on the UK pregnancy register. Jayne Fotheringham, chief technician, will discuss the use and interpretation of EEG in epilepsy. Dr Leslie Mac, paediatric neurologist, will talk on the subject of MRI.

You will have the choice of listening to specialists in paediatrics, adults and learning disabilities on the topic of 'It's not always epilepsy'.

An afternoon on 'How to get the best from you and your patient' will be led by Dr John Paul Leach, consultant neurologist. As well as sharing his clinical experience of working with people with epilepsy there will be workshops in the three specialist areas. Finally there will be the ESNA Annual General Meeting and presentation of the Malcolm Taylor Award for the best poster presentation and the Achievements in the Care of Epilepsy Awards.

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