

Sixth WMS Satellite teaching course: 28th-29th September 2008

Teaching objective: To improve the diagnostic competence of professionals dealing with patients with neuromuscular diseases.

Target audience: medical specialists (paediatricians, neurologists, paediatric neurologists, geneticists, neuropathologists) and medical specialists in training, and also other attendees of the WMS congress.

Rationale

There is a general feeling that diagnosing neuromuscular diseases is cumbersome. This is mainly due to the fact that there are hundreds of different neuromuscular diseases, and in addition each disease in itself is rare. However, taken together the number of patients with neuromuscular disorders is considerable. Different specialists are confronted with patients suffering from a neuromuscular disorder during clinical practice. Neuromuscular disorders may occur in newborns, but also develop at an advanced age. They can have a hereditary origin, an auto-immune pathogenesis, a neurodegenerative nature or they can occur as a complication of a systemic disease. Diagnosis of neuromuscular diseases requires special skills. It all starts with asking the right questions (clinical and genetic history taking) and performing a thorough clinical examination including manual muscle testing and functional tests. History taking and clinical examination form the basis for additional ancillary investigations, which might include electrophysiological studies, muscle or nerve biopsy, genetic testing, muscle imaging, metabolic studies, etc. During higher specialist training it is usual that little attention is paid to neuromuscular disorders. Due to general lack of familiarity with this group of diseases misdiagnosis is not infrequent. It is of utmost importance to establish a correct diagnosis, not only because sometimes neuromuscular diseases are treatable, but also because a precise diagnosis is required for prognostication and genetic counselling. This course provides a context to develop these skills through discussion of patients and review of biopsies. Feedback from the first five courses (held from 2003 onwards) was overwhelmingly positive.

Attendants: max. 40

Venue:

Bioscience Centre, International Centre for Life and Royal Victoria Infirmary, Newcastle upon Tyne

Cost:

£200 + VAT (£235) without accommodation

£340 + VAT (£399.50) with 2 nights accommodation at Express by Holiday Inn

Cost includes course fees, 2 dinners, 1 lunch and mid morning/afternoon refreshments

Full booking form will be available early April, you may pre-register now.

An Integrated View on Neuromuscular Disease

Course description:

Recently, there have been major advances in our understanding of the pathophysiology, molecular genetics and treatment of a good deal of neuromuscular diseases. These discoveries have had a major impact on our ability to establish an accurate diagnosis, on genetic counselling and on our management of these disorders.

The emphasis of this course will be on the clinical approach to the evaluation of patients who present with symptoms of weakness and the interpretation of the muscle biopsy.

The course will be entirely practical and case reports will be presented to highlight key clinical concepts.

There will be the opportunity to discuss the diagnostic approach to patients shown on video, and to look at muscle biopsies. Generous time will be allowed for questions from the audience, and for discussion of difficult topics.

Participants will be encouraged to bring difficult, unusual or unsolved cases for discussion.

For this purpose, each participant will be asked to present one neuromuscular case to be presented and discussed.

For every case he/she should prepare:

- 1) Two Powerpoint slides
 - Slide 1 should contain: a clinical picture/ clinical video demonstrating major clinical characteristics (onset; clinical course; CK; NCV; heredity; others);
 - Slide 2 should contain: one pertinent piece of information (e.g. muscle or brain imaging; a family tree; immunofluorescence or Western blot data)

This presentation should not take longer than 3 minutes.

- 2) Bring along a maximum of 5 biopsy slides (obligatory: HE, Trichrome plus 3 optional) to be viewed under the microscope and discussed.
- 3) **A clinical vignette of this case must be submitted 4 weeks ahead of the course to the course organizers.** The vignettes will be circulated.

Upon completion:

Participants should be able to better recognise a number of important neuromuscular presentations; incorporate up-to-date diagnostic approaches to the patient suspected of a neuromuscular disease; consider the differential diagnosis; apply appropriate tests.

The focus of the course will be on interaction between faculty and course attendants, and on obtaining hands-on experience with clinical examination and reading muscle biopsies.

Sixth WMS Satellite Teaching Course

(Proposed Programme, please note this may be subject to change)

Saturday September 27th 2008

Arrival of delegates

Delegate to meet at the Express by Holiday Inn at 7pm before going on to an evening meal, restaurant to be confirmed.

Sunday September 28th

8.00 Coffee and bacon rolls available at registration, Bioscience Centre, International Centre for Life

8.45 What can we learn from the clinical and genetic history and how do we perform a clinical examination on adults – hereditary diseases? – Kate Bushby

9.30 What can we learn from the clinical and genetic history and how do we perform a clinical examination on adults – acquired diseases? – David Hilton-Jones

10.15 How do we perform a clinical examination on children? – Carsten Bonnemann

11.00 Coffee break

11.30 What can we learn from the muscle biopsy, part 1: light microscopy of hereditary diseases – Caroline Sewry

12.00 What can we learn from the muscle biopsy, part 2: light microscopy of acquired diseases and electronmicroscopy – Anders Oldfors

12.30 Molecular diagnostic testing and what does it mean? – Volker Straub

13.00 Lunch

15 minute Walk to the Royal Victoria Infirmary for afternoon session - group split into two (group A and group B)

14.15 Group A- guide to muscle examination in adults and children and opportunity to meet patients with different diagnoses

Group B- muscle pathology session in Histopathology suite

16.15 Coffee

16.30 Group A- pathology session in Histopathology suite

Group B- guide to muscle examination in adults and children and opportunity to meet patients with different diagnoses

18.30 Close of session walk to meal at Barn Asia (next to Express by Holiday Inn)

Monday September 28th

- 8.00 Coffee and bacon rolls available at Bioscience Centre, Centre for life
- 8.45 Systematic overview on the floppy infant presentation, and diagnostic approach – Thomas Voit
- 9.45 Systematic overview on patients presenting with limb-girdle and distal muscle weakness, and diagnostic approach – Marianne de Visser
- 10.45 Coffee
- 11.00 Presentation of difficult or unsolved cases – participants
- 13.00 End of the meeting