Report on the 12th and 13th Asian and Oceanian Myology Center (AOMC) Annual Scientific Meetings

The Asian and Oceanian Myology Center (AOMC) was established as an organisation in Tokyo in 2001. Its aims and objectives have been to promote scientific and clinical research, to provide a forum to advance and distribute knowledge for the benefit of patients, to provide educational opportunities for young investigators and clinicians and to promote the achievement of high standards in clinical practice in myology and neurology as they relate to neuromuscular diseases in the Asian and Oceanian region.

12th Meeting (June 6-8, 2013 Xi'an China)

For this meeting, the program committee including the honorary chairman, Professor Dingguo Shen and his colleagues, with the assistance of AOMC Executive Board Members, selected the following topics including

1. **Inflammatory myopathies**

   Inclusion body myositis (IBM), childhood dermatomyositis and mitochondrial changes in IBM

2. **Metabolic myopathies**

   Mitochondrial respiratory chain disorders, lipid storage myopathies and Pompé disease (newborn screening and enzyme therapy)

3. **Therapeutic trials and management of muscular dystrophy**

   Exon skipping approach for Duchenne muscular dystrophy (DMD), AAV-mediated DMD gene replacement therapy and management of muscular dystrophy patients

4. **Diagnostic techniques for neuromuscular diseases**

   Muscle imaging (MRI) of DMD patients, muscle channelopathies and electrophysiological investigation

5. **Myopathy with GNE gene mutations**

   Sialic acid trials as a therapy for GNE myopathy and experience of GNE myopathy in China, Thailand, Malaysia and Korea

6. **Clinicopathological conference (CPC) regarding 10 patients**

   As usual, there was a Muscle Pathology Case Conference during which 10 of the most interesting and unusual cases were presented after being chosen by the conference organisers from many submitted. Sets of original microscope slides relating to these cases were available for viewing throughout the meeting. These sessions are invariably greatly appreciated and enthusiastically received by both novices and experienced researchers, frequently with extensive discussion and divergent opinions before reaching a final diagnosis or concluding that further information/biochemistry/genetic analysis is required.

Through the presentations and discussion during this conference, we were particularly impressed with the following revelations unique to Asian countries:

Patients with inclusion body myositis (IBM) are quite rare in South Asian countries but there are increasing numbers in Japan

The incidence of IBM has been markedly increasing during the most recent 10 years in Japan. It has now reached almost the same level as in Western countries. Since the number of patients with IBM is very low in South Asian countries, this may give us

some hints towards clarifying the pathogenic mechanism of the disease, especially if we were to undertake a comparative study of HLA typing, in conjunction with changes in life style, diet and environment.

(2) Lipid storage disease is common, especially in the southern part of China

Professor Chuanzhu Yan reported that one of the lipid storage myopathies, multiple Acyl-CoA deficiency (MADD) is common in the southern part of China and is mostly treatable with riboflavin. The most common mutation is c250G>A. On the other hand, patients with MADD are quite rare in Japan and other Asian countries. At any rate, it is exciting news that many lipid storage myopathy patients are recovering after riboflavin administration in China.

Overall, the meeting was very successful with 188 abstracts also submitted for the interesting poster presentations that covered most areas of Neuromuscular Disease.

13th Meeting (May 14-17, 2014 Makati City Philippines)

This AOMC meeting was co-organized with the 20th Philippine Neurological Association (PNA) midyear convention on May 14-17 in Makati city (a satellite city of Manila) by AOMC Vice President, Dr Raymond L. Rosales. With two distinguished guests, Professor Hans H. Goebel (Mainz, Germany) who discussed ceroid lipofuscinosis and Professor Bjarne Udd (Vasa, Finland) whose presentation was on distal myopathy, 400 participants including 40 foreign delegates considered the following topics. Since many young neurologists were attending, who would be unfamiliar with neuromuscular disorders, educational courses in clinico-pathological, electrophysiological and neuro-imaging diagnostics were planned. The plenary sessions included:

1. **Muscular dystrophy**

   Updates in the diagnosis of facioscapulohumeral muscular dystrophy (FSHD), gene therapy in Duchenne Muscular Dystrophy (DMD) and dystrophinopathy in female patients.

2. **Mitochondrial and metabolic myopathies**

   An overview of the clinico-pathological spectrum of lipid storage myopathies, newborn screening for Pompé disease (the Asian experience) and pharmacological therapy of mitochondrial myopathies.

3. **Channelopathy and toxin myopathy**

   Electrophysiological diagnosis of muscle ion channelopathies, pathophysiology of primary hypokalemic periodic paralysis (HOKPP) and statin-associated myopathy.

4. **Treatment strategies in neuromuscular diseases**

   Critical and respiratory care in neuromuscular disorders, Rituximab in refractory autoimmune myositis and stem cell therapy in neuromuscular disorders.

5. **Clinico-pathological conference regarding 10 patients**

   Once again, an interesting and unusual set of cases was presented. Relevant histopathological slides had again been available, for viewing by all those interested, throughout the meeting. These cases generated considerable thought and a good deal of discussion.

Through the presentations and discussion during this conference, we were particularly impressed with the following revelations that seem fairly unique to Asian countries:

(1) Treatment for metabolic myopathies

At the last AOMC meeting in Xi'an, Dr Chuanzhu Yan emphasized the observation that one of the lipid storage myopathies, multiple Acyl-CoA deficiency (MADD) with the c250G>A mutation, had been found to be common in the southern part of China and, importantly, that most patients proved treatable with riboflavin (Vitamin B12) administration. Dr Nishino also emphasized that MADD is treatable, but that the incidence is far less frequent in Japan. He also described three patients with primary carnitine deficiency who were treated with oral carnitine administration.

For several years now, newborn screening for Pompé disease has been performed throughout Taiwan using dried blood spots with the incidence of the disease being estimated to be 6/100,000. This has enabled earlier enzyme replacement therapy (ERT) resulting in excellent recovery towards normal muscle function. Incidence of the disease in other Asian countries remains unknown. Unfortunately, enzyme replacement therapy is almost impossible in most Asian countries because of the expense involved.

(2) Treatment and care management for Duchenne muscular dystrophy

There appears to be an increasing hope of treating DMD patients through exon skipping therapy and stem cell therapy, although exon skipping has, so far, failed to demonstrate unequivocal effectiveness in double blind clinical trials. Drs Takeda and Khadilkar discussed advances in stem cell therapy, though there are still many major barriers to overcome before muscle stem cell therapy can be applied to patients. With advanced patient care, the average life-span of young men with DMD has increased from 18 years to over 30 years. The main factors leading to the prolonged life span are improvements in cardiac management and respiratory care by using non-invasive positive pressure ventilation (NPPV), but not all patients can have the same care management because

support systems differ from country to country. We need further collaboration to standardize disease treatment and care across our region.

This meeting was also very successful, with all participants learning more about the diagnostic methods that can lead to treatment. There is considerable hope that ongoing collaboration between Asian and Oceanian countries will have a significant effect on improving the treatment of all our patients with neuromuscular diseases.

The 14th AOMC meeting will be held on March 1-4 in Bangkok, Thailand.

AOMC (https://www.aomc.info/)

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