



**Service Priorities and Programmes**  
**Electronic Presentations**

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**The Comprehensive Neuromuscular Diagnostic Program in Hong Kong West Cluster**

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**Introduction**

There are many different types of paediatric neuromuscular disorder. While diagnosis of some rare neuromuscular disorder can be difficult, a team of specialists is required to provide state-of-art-diagnostic evaluation in a timely manner. The neuromuscular diagnostic team include paediatric neurologists, neuropathologists, chemical pathologists and radiologists with special expertise in the paediatric neuromuscular field. Since 2009, the neuromuscular diagnostic clinic was established with a team of specialists regularly meeting together. We accept referral from all the HA hospitals and all the patients that had been referred to this clinic were cases of challenging diagnosis. All patients discussed at the clinic had received detail evaluation, investigation studies, electromyography and muscle biopsy, and some had involved overseas collaboration during the diagnostic process. Diagnostic and management plan were decided at the meeting for each patient.

**Objectives**

This study is to evaluate the effectiveness of this neuromuscular diagnostic program involving a multi-specialty team approach, and also to report the development of this service.

**Methodology**

To review the number of neuromuscular disorder cases that had been reviewed at the 12 neuromuscular diagnostic clinic sessions and the diagnoses of the different rare neuromuscular disorders that had been confirmed.

**Result**

From September 2009 to February 2014, there were a total of 12 neuromuscular diagnostic clinic sessions which involved a multi-specialty team approach. A total of 31 paediatric patients were evaluated by the team. Specific diagnosis could be

concluded in 26 patients including different subtypes of congenital myopathy, muscular dystrophy, and other rare conditions such as Pompe disease and Danon Disease. The immunohistochemical staining panel was established in 2012 and 2013 in two phases. The diagnostic MRI muscles imaging were introduced in 2013. The confirmed diagnosis enables initiation of timely treatment, appropriate referral of rehabilitation services, and provision of suitable genetic counseling and prognosis guidance. While paediatric neuromuscular disorder is a group of heterogeneous, rare and diagnostic challenging conditions, our multi-specialty team approach has been shown to be an effective first step in patient management.