Diagnosis and Treatment of Neuromuscular Diseases – Local Advancement and Challenges

Abstract

Paediatric neuromuscular diseases (NMDs) describes a group of hereditary condition associated with disorders in the spinal motor neurons, peripheral nerves, neuromuscular junction and muscles, that often lead to muscle weakness, motor difficulties and sometimes other systems are also involved. Some of these conditions could have a progressive deteriorating course. In this review, an update regarding the most common neuromuscular diseases like spinal muscular atrophy and Duchenne muscular dystrophy, as well as the rare hereditary NMDs including non-5q SMA, congenital muscular dystrophy, congenital myopathy and congenital myasthenic syndrome will be discussed focusing on the contemporary approach to diagnosis and treatment in our locality.

Several promising therapeutics are now being actively tested in clinical trials for some common neuromuscular diseases with the current better understanding of the underlying genetic pathogenic mechanism causing the important protein loss or dysfunction. While we are actively participating in some of these internationally collaborated trials, the local patient registry, the standardization of care and the closely collaborated teamwork allows the early identification and recruitment of the suitable patients. With the recent overseas approval of some of the expensive treatment, we will also face a new era of management for our patients and families , with new challenges too very soon.

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