Importance of Timely and Accurate Diagnosis of Myotonic Disorders: Role of Electromyography

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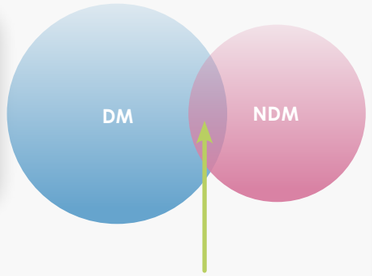
Myotonic Dystrophy and Non-dystrophic Myotonias

- Myotonic disorders are a heterogeneous group of interited neuromuscular disorders[1]
- Myotonia is a symptom that is a common feature of several types and subtypes of myotonic disorders, including myotonic dystrophy (DM) and non-dystrophic (NDM) myotonias.
- Myotonia presents clinically as delayed muscle relaxation after voluntary contraction, leading to muscle stiffness or cramping, and/or electrophysiologically as spontaneous discharge of muscle fibres[1]

DM subtypes: DM1; DM2

Prevalence: 1 in 8,000 for DM1; DM2 rarer[2,3]

Diagnostic delay: typically >5 years (DM1) and >14 years (DM2)[2]



NDM subtypes:
Na^+ channelopathy;
Cl^- channelopathy

Prevalence: 1 in 250,000 to 1 in 400,000 for autosomal dominant forms; 1 in 25,000 for autosomal recessive forms[3]

Diagnostic delay: typically >10 years[4]

Myotonia is the main symptom of NDM, and one of many possible symptoms of DM

Diagnosis of Myotonic Disorders

Diagnostic Pathway *(adapted from Stunnenberg et al 2020[5])*

Key information:
- Age of onset
- Family history
- Symptoms
- Exacerbating factors

Key initial diagnostic test:
- Neurological examination
- Bedside test of motor function
- Observation of stiffness level

Recommendation that patients fill out a symptom diary

Follow-up tests:
- Electromyography (EMG)
- Genetic testing

Diagnostic delays

Variable, non-specific symptoms[4-7]
- Muscles affected
- Disease severity
- Age of onset
- Warm-up phenomenon
- Cold phenomenon
- Overlap with other diseases

Coping[8]
- A "family problem" (inherited disease) people manage by themselves

There are numerous reasons for delays in diagnosing DM and NDM

Lack of disease recognition[4]
- Rare diseases, not frequently encountered by non-specialist HCPs

Delays seeking medical help[9]
- Patients don't ask
- Non-specialists don't refer on

Under-recognised disease burden[4]
- Underestimation of impact on patients and carers

- Diagnostic delays have a negative impact on patients' wellbeing,[2] as they have to learn to cope with their condition, often by limiting what they do, instead of being offered treatment to ameliorate symptoms
- Timely and accurate diagnosis is important for genetic counselling and screening of systemic features in DM, as well as determining appropriate management[1]
- Using EMG can help provide timely confirmation of a diagnosis

Role of EMG in DM

Low need for EMG when there is a clear DM phenotype and clear signs of clinical myotonia

Greater need for EMG when:

Genetic screening is required

Myotonia fluctuates or is difficult to find

Role of EMG in NDM

Useful when clinical myotonia is uncertain

Helpful when interpreting relevance of a gene variant, and ensuring correct diagnosis

Can prevent delay to appropriate medical management

Can prevent erroneously offering inappropriate medical management

Key Learnings

- EMG, while not necessary in every case, plays an important role in the timely diagnosis of both DM and NDM
- EMG, alongside genetic testing, can facilitate accurate differential diagnosis of disease subtypes

Abbreviations
DM: myotonic dystrophy; EMG: electromyography; HCP: healthcare professional; NDM: non-dystrophic myotonia.

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