

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

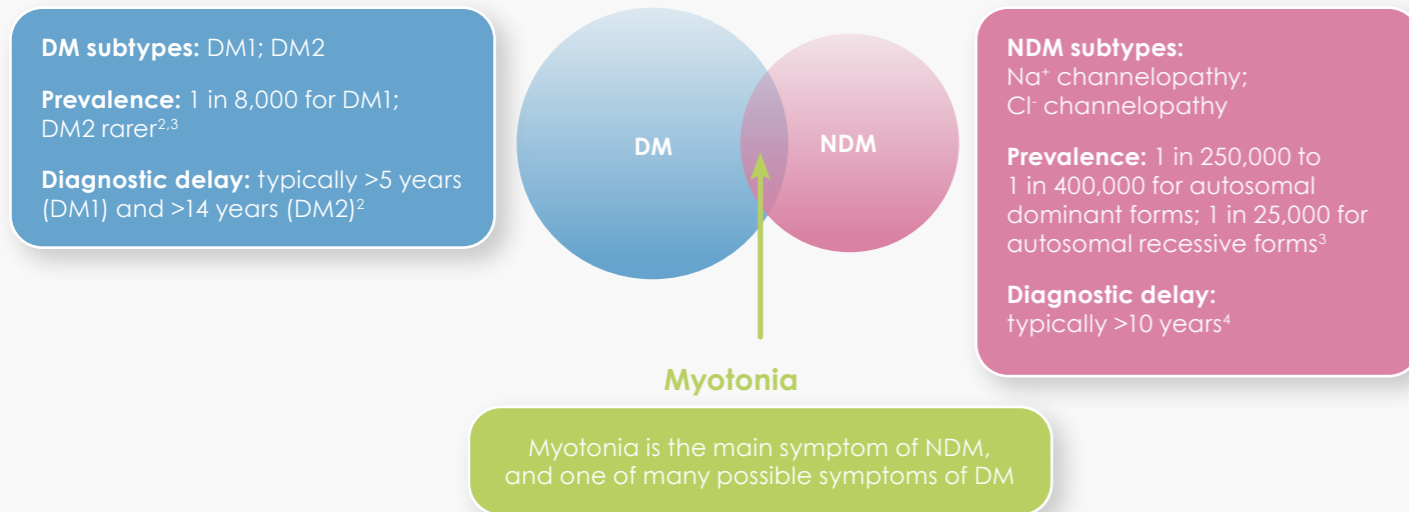
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## Myotonic Dystrophy and Non-dystrophic Myotonias

- Myotonic disorders are a heterogeneous group of inherited neuromuscular disorders<sup>1</sup>
- 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<sup>1</sup>



## Diagnostic delays

### Variable, non-specific symptoms<sup>4-7</sup>

- Muscles affected
- Disease severity
- Age of onset
- Warm-up phenomenon
- Cold phenomenon
- Overlap with other diseases

### Coping<sup>8</sup>

- A "family problem" (inherited disease) people manage by themselves

### There are numerous reasons for delays in diagnosing DM and NDM

### Lack of disease recognition<sup>4</sup>

- Rare diseases, not frequently encountered by non-specialist HCPs

### Delays seeking medical help<sup>9</sup>

- Patients don't ask
- Non-specialists don't refer on

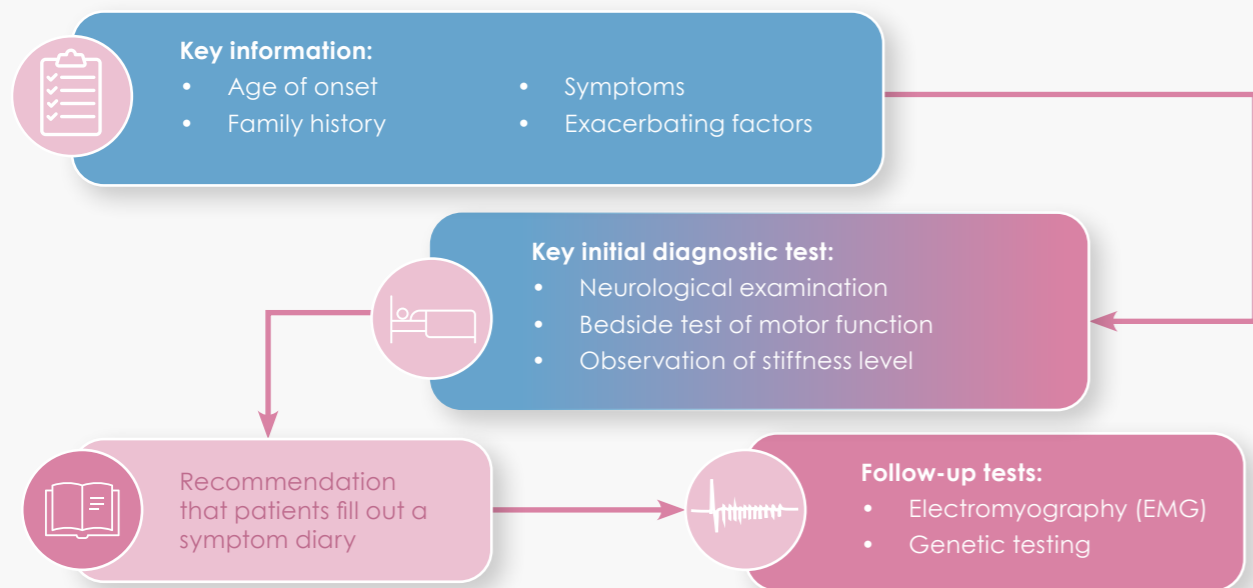
### Under-recognised disease burden<sup>4</sup>

- Underestimation of impact on patients and carers

- Diagnostic delays have a negative impact on patients' wellbeing,<sup>2</sup> 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<sup>1</sup>
- Using EMG can help provide timely confirmation of a diagnosis

## Diagnosis of Myotonic Disorders

Diagnostic Pathway (adapted from Stunnenberg et al 2020<sup>5</sup>)



## 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

### Abbreviations

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

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## 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