Myotonia: Are We Underestimating the Burden in Myotonic Disorders?

Raising awareness of the burden of disease due to myotonia will enhance understanding of patients' needs, and improve the management of myotonic disorders.

This infographic presents highlights from a symposium held on 23rd April 2024 as part of the 8th International Myology Conference in Paris, France. The symposium and infographic were sponsored by Lupin Neurosciences.

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Myotonic Disorders Are Rare Inherited Neuromuscular Diseases[1-4]

Myotonic Dystrophy (DM)[1,2]

- Rare (AD inheritance; DM has a global prevalence of 1 per 10,000[5])
- Impaired Cl^- channel function
- **Multisystemic: Myotonia is one of many possible symptoms**
- DM2 symptoms can be non-specific and replicate other conditions

DM1
- Congenital, childhood, and adult onset

DM2
- More commonly adult onset

Non-Dystrophic Myotonia (NDM)[3,4]

- Ultra rare (prevalence AD forms: 1 in 250,000 to 1 in 400,000; AR forms: 1 in 25,000)
- Impaired Cl^- or Na^+ channel function
- **Myotonia is the main symtom[6]**
- Variable age of onset, usually presents in first 2 decades of life

Cl^- channelopathy
- Becker MC (AR inheritance)
- Thomsen myotonia congenita (MC)

Na^+ channelopathy
- Paramyotonia congenita
- Sodium channel myotonia*
- Closely related *SCNC4A* myotonic disorders

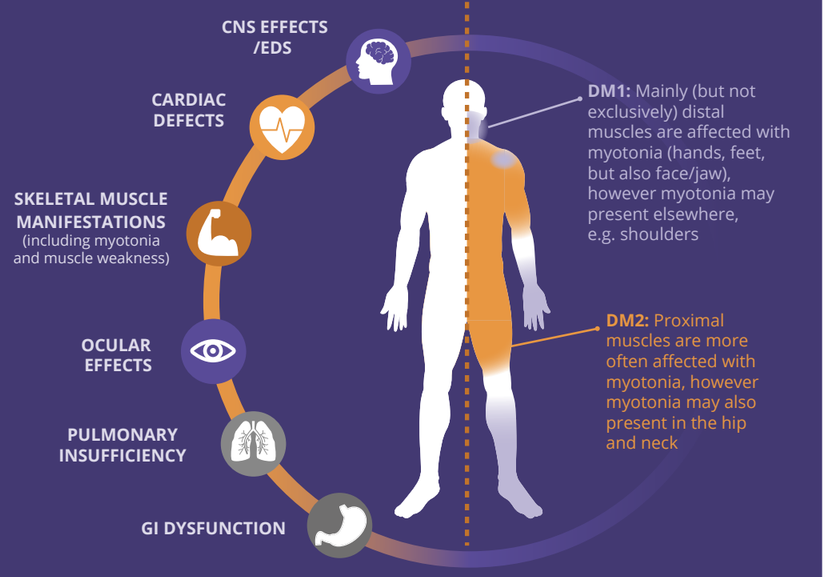
*Includes myotonia fluctuans, myotonia permanens, and acetazolamide-sensitive myotonia.

DM1 Has Multisystemic Manifestations[1,7-14]

Multiple symptoms manifest at different ages and in all forms of DM1 onset.[15,16]

Location of Myotonia in DM1 and DM2[15,16]

Myotonia precedes all other symptoms (including muscle weakness), and is present in all age groups, apart from at birth.[15,16]



Myotonia Symptoms Affect Many Different Areas of Patients with Myotonic Disorders' Daily Lives[1,6-9,16-20]

The ENSA patient survey showed that 93% of patients had a current or previous history of myotonia; however, the burden of disease due to myotonia has not been fully addressed in these patients.[21] Disease burden due to myotonia may include the following:

Unpredictable attacks
Attacks are life-long, unpredictable, and disabling.
Feeling restricted, persistent tiredness, pain, and cramping.

Social anxiety
Unable to release handshake.
Difficulty eating.
Problems speaking.

Issues with daily living
Challenges with dressing.
Difficulty with personal hygiene.

Reduced activity
Difficulty exercising or climbing stairs.
Work duties can be challenging.
Difficulty crossing streets.

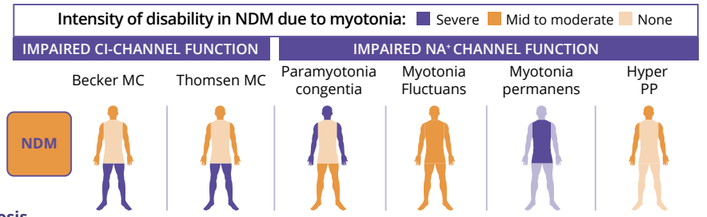
Lack of independence
Occasional assistance if needing to drive.
Need help with some tasks.

Myotonia e.g. Grip Myotonia Is a Common, Overlapping Symptom in Myotonic Disorders that Impacts Quality of Life

- muscle stiffness/delayed muscle relaxation after forced contraction[4,22]
- impacts quality of life of individuals with myotonic disorders, including DM and NDM[6,8,9,18,23]
- is often perceived as lower priority versus other symptoms in individuals with multisystem myotonic disorders like DM[9]

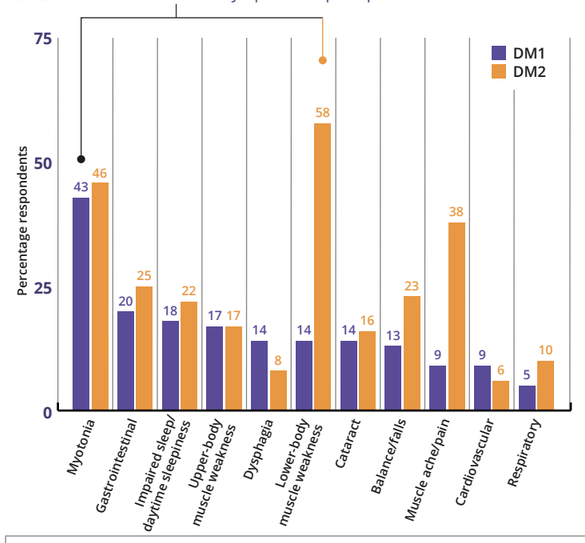
Delayed Diagnosis of DM and NDM

- Diverse genetic and phenotypic manifestations make diagnosis of DM and NDM challenging[8]
- DM diagnosis is typically delayed by 3–12 years[24]
- NDM diagnosis is typically delayed by 15–17 years[6,25]
- The **IMPACT** patient survey showed that **65% of patients** had **NDM** symptoms for >10 years before diagnosis[6]
- **Myotonia intensity varies in NDM, depending on diagnosis**



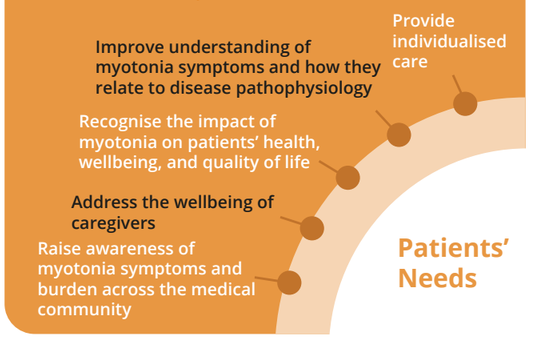
Living with Myotonia: Understanding the DM Patient Experience

Myotonia in patients with DM1 and lower body muscle weakness in patients with DM2 are the most common symptoms to prompt a first clinical consultation[21]



Objectives to Improve the Clinical Journey for Patients with Myotonia

Myotonia is one of the top five symptoms that patients with DM want to improve[21]



Abbreviations
AD: autosomal-dominant; AR: autosomal-recessive; Cl^-: chloride ion; CNS: central nervous system; DM (1/2): myotonic dystrophy (Type 1/2); EDS: excessive daytime sleepiness; EMG: electromyography; GI: gastrointestinal; hyper PP: hyperkalemic periodic paralysis; MC: myotonia congenita; Na^+: sodium ion; NDM: non-dystrophic myotonia.

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