Diagnostic Odyssey of Myotonic Disorders

Insights from a patient and caregiver survey, treating physicians and scientific literature.

Patient/caregiver surveys:
- 39 Participants with Myotonic Disorders
  - 19 NDM with chloride channelopathies
  - 7 NDM with sodium channelopathies
  - 10 DMI
  - 2 DMO

KOL roundtable to leverage insights from survey participants with experience in clinical practice:
- 7 pediatric neurologists
- 2 adult neurologists

Exports from:
- Belgium
- Spain
- Italy
- Germany
- France

NDM paediatric patient journey

DM paediatric patient journey

Present from birth with main symptoms being severe generalized weakness, hypertonia and respiratory compromise. Myotonia is usually absent in infancy, and muscle strength can improve with time if the infant survives.

Key
- Symptoms
- Diagnosis
- Treatment
- Barriers

DMN C2–channelopathy: 12-18 Palsy of Age
Conceived prevalence — 1:100,000
- DMC: ~25,000,000
- TRMC: ~2,000,000

Predominant symptoms
- Limb stiffness (limb-limb more often)
- Grip myotonia
- Pain with stiffness
- Coldtrigger
- Warm-up phenomenon

DMN A–channelopathy: 2-6 Years of Age
Conceived prevalence — 1:100,000
- PMC: ~250,000
- HyperPP: ~1,000,000

Predominant symptoms
- Pain with stiffness
- Episodic weakness
- Grip myotonia
- Coldtrigger
- Eye closure myotonia
- Limit stiffness
- Facial stiffness
- SMEx, hypertonia and laryngospasm

DMN: 13-15 Years of Age
DMC: Estimated prevalence of 3-10,000,000 (higher prevalence in Individuals with specific genotypes). Myotonia and weakness tend to worsen in late childhood and adolescence.

Myotonia in DMC is considered a secondary symptom, and is underestimated by HCPs.

NMD paediatric patient journey

Birth

C1 channelopathy: Symptoms may be present from birth.

1 Year

Congenital DMI: Myotonia symptoms may start to become apparent.

DIAGNOSIS

3 Year

Elucidation myotonia: Extent within the first decade of life.

5 Year

C1 channelopathy: Symptoms usually present in the first decade of life with stiffness in legs more than arms, hands and face.

9 Year

C1 channelopathy: Symptoms may become apparent when children begin to participate in sports.

IMC myotonia: Onset from 4-22 years of age. Hypertonia myotonia (onset from 2-30 years of age.

10 Year

Myotonia treatment age: ≥10 years.

No curative treatment, off-label symptomatic treatment include azacitidine, carbamazepine, flecainide, mesoline.

12 Year

Diagnosis difficult in patients with no family history.

13 Year

Diagnosis can be delayed up to 18 years. Age of diagnosis is variable, but often is early adolescence.

Embarrassment may prevent children seeking advice about their symptoms.

DM: Myotonic symptoms can present from 6 years, but are more commonly seen from late adolescence.

Cardiac arrhythmias or other cardiac conditions may occur as patients enter adulthood.

18 Year

Access to treatment can be hindered by lack of awareness of poverty and impact of myotonia.

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Diagnosis of DMI typically occurs in adulthood.

17 Year

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4 Year

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3 Year

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2 Year

Diagnosis of DMI typically occurs in adulthood.

1 Year

Diagnosis of DMI typically occurs in adulthood.

0 Year

Diagnosis of DMI typically occurs in adulthood.