

Diagnostic Odyssey of Myotonic Disorders

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

Patient/caregiver surveys:



KOL roundtable to leverage insights from survey participants with experience in clinical practice:



Key

- Symptoms
- Treatment
- Diagnosis
- Barriers

NDM paediatric patient journey

DM paediatric patient journey



NDM Cl⁻ Channelopathy: 13-18 Years of Age

Combined prevalence = ~1:100,000 (higher in Scandinavia (7-10:100,000))

- BMC = ~1:250,000
- TMC = ~1:400,000

Predominant symptoms

- Limb stiffness (lower limbs more often)
- Grip myotonia
- Pain with stiffness
- Cold trigger
- Warm-up phenomenon

NDM Na⁺ Channelopathy: 13-18 Years of Age

Combined prevalence = ~1:100,000

- PMC = ~1:250,000
- HyperPP = ~1:200,000

Predominant symptoms

- Pain with stiffness
- Episodic weakness
- Grip myotonia
- Cold trigger
- Eye closing myotonia
- Limb stiffness
- Facial stiffness
- SNEL: hypertonia and laryngospasm

DM: 13-18 Years of Age

DM1: Estimated prevalence of 3-15:100,000 (higher prevalence (≥20X) in Sweden, Basque of Spain and Quebec)

Childhood-onset DM1: no published data

Congenital DM1: exact global prevalence is unknown; however, congenital DM1 likely represents 10-30% of overall DM1 population

Estimates of incidence:

2-28:100,000 live births

DM2: prevalence unknown