

Development of an auto-questionnaire to measure the impact of non-dystrophic myotonia on patients' daily living and quality of life

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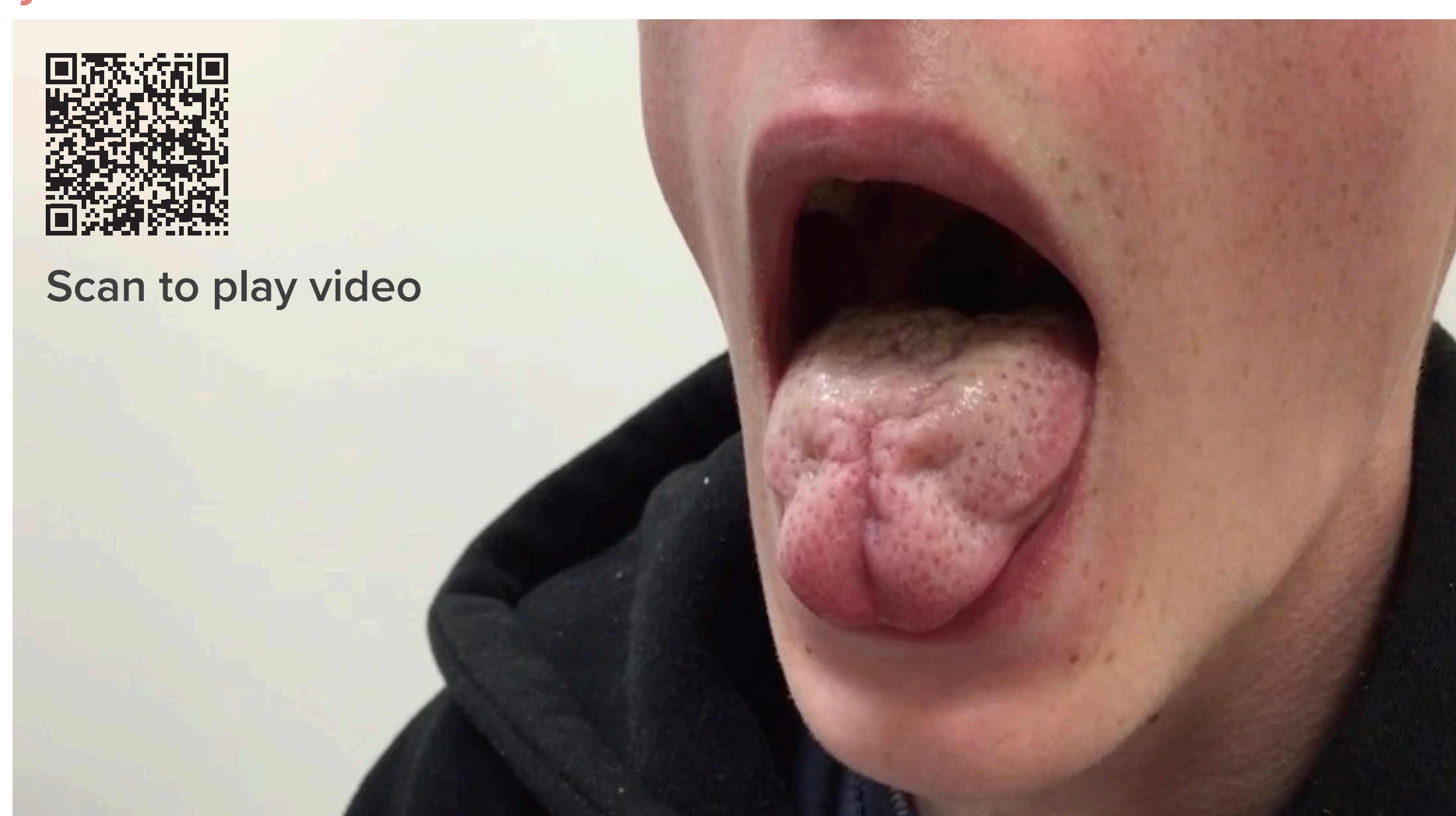
Introduction and rationale

- Symptoms of the non-dystrophic myotonias (NDM) vary considerably depending on the disease subtype, and differ in frequency and impact between individuals.¹⁻³
- Understanding the repercussions of NDM on daily life and quality of life (QoL) is pivotal for treatment planning, clinical management and monitoring.
- A number of tools to assess the impact of NDM on a patient's life have developed and validated. However, no NDM-specific tools provide standardised, reproducible, patient-reported assessments across the varied symptoms experienced with NDM.^{4,5}

Hand grip myotonia



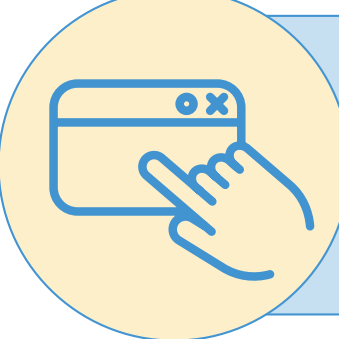
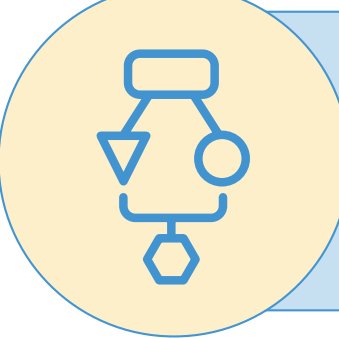
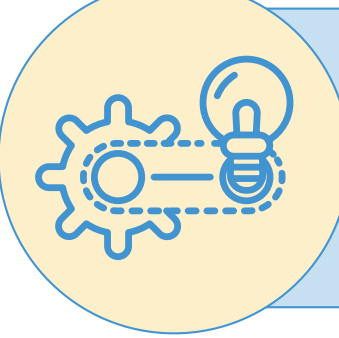


Tongue myotonia



Objectives

- To develop a questionnaire that evaluates the personal impact of NDM, for completion by patients during routine consultations.
- This questionnaire aims to highlight key areas necessitating clinical attention:
 - It aims to be a valuable tool to support diagnostic and follow-up consultations.
 - It also aims to provide patient-reported outcomes (PROs) that facilitate informed clinical decisions.

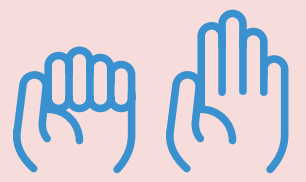
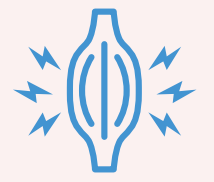
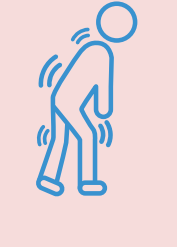

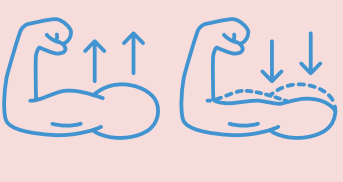




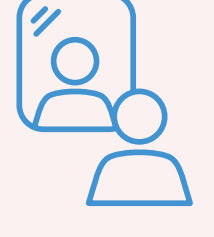
Methods

-  Literature review of 5 studies⁶⁻⁹ investigating NDM symptoms and management, to identify domains with greatest impact on daily living/quality of life
-  Items within these domains were classified by mapping them against validated tools that explore physical and mental health in NDM and other diseases
-  Prototype questionnaire developed/discussed by French experts, then tested in people with NDM by the multidisciplinary team at CHU Nice
-  CHU Nice team will revise the questionnaire based on patient validation work and clinical-practice feedback in pre- and post-questionnaire settings
-  Questionnaire to be revised and its value established at international level

Results

- The literature review identified 10 domains with greatest impact on daily living/quality of life in people with NDM:
 - Myotonia; pain; muscle weakness; fatigue; atrophy/hypertrophy; activity; emotions; independence; relationships; body image.
- These domains were mapped against validated clinical tools (Figure 1).
- The first version of the questionnaire contained 100 items:
 - These items were revised and validated at an advisory board meeting attended by nine French NDM experts, resulting in the second version (36 items).
- A protocol has been drawn up and agreement has been obtained to conduct a questionnaire validation study, enrolling 10 patients with NDM at CHU Nice.

Figure 1: In the development process for the auto-questionnaire, 10 domains with greatest impact on daily living/QoL in NDM were mapped against clinically validated tools

1. Myotonia		<ul style="list-style-type: none"> • MDHI Myotonic Dystrophy Health Index • NDMRS: Non-dystrophic Myotonia Rating Scale • MEF: Myotonia Evaluation Form
2. Pain		<ul style="list-style-type: none"> • MPQ: McGill Pain Questionnaire • HNPQ: Hereditary Neuropathy Pain Questionnaire
3. Muscular weakness		<ul style="list-style-type: none"> • NMAS: Non-Dystrophic Myotonia Activity Scale • MDHI: Myotonic Dystrophy Health Index
4. Fatigue		<ul style="list-style-type: none"> • FSS: Fatigue Promis
5. Atrophy/Hypertrophy		<ul style="list-style-type: none"> • MAFT: Muscle Atrophy Focused Test • MHFT: Muscle Hypertrophy Focused Test
6. Activity		<ul style="list-style-type: none"> • SF36 • DM1-Active-1c • Becker physical activity questionnaire • Physical activity questionnaire from the Michael J. Fox Foundation for Parkinson's Research • Women's Health Initiative physical activity questionnaire
7. Emotions		<ul style="list-style-type: none"> • SMDMHQ: Steinert Myotonic Dystrophy Mental Health Questionnaire • SF36 • HADS: Hospital Anxiety and Depression Scale
8. Independence		<ul style="list-style-type: none"> • Barthel Index • Lawton and Brody Index • NMAS: Non-Dystrophic Myotonia Activity Scale
9. Relationships		<ul style="list-style-type: none"> • SF36 • SMDMHQ: Steinert Myotonic Dystrophy Mental Health Questionnaire
10. Body image		<ul style="list-style-type: none"> • BIDQ: Body Image Disturbance questionnaire • BDDE Body Dysmorphic Disorder Examination

Conclusions

- Although establishing the impact of NDM on QoL is important for treatment planning/monitoring, no disease-specific tools provide standardised, reproducible, PRO assessments across the spectrum of NDM symptoms.
- An questionnaire is being developed by the multidisciplinary team at the Rare Neuromuscular Disease Reference Center, CHU Nice, to measure the impact of NDM on patients' daily living and QoL.
- The questionnaire aims to be a robust NDM-specific tool that establishes the frequency and impact of key symptoms, emphasises aspects of clinical priority, facilitates consistent PRO assessment of NDM progression and evaluates treatment.
- The questionnaire testing and validation process, involving people with NDM and international multidisciplinary clinical experts, aims to determine the useability and value of this tool.

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Disclosures

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