admedicum 10 years
PATIENT DRIVEN SOLUTIONS

6 5 4 3 2 1

Inspiring Anniversary Highlights

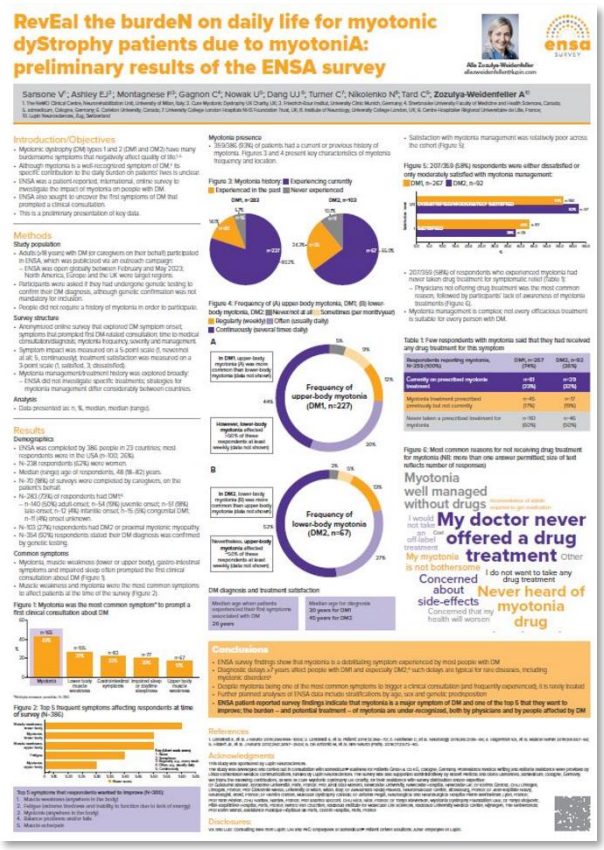
Co-creating Patient Experience Data:
Insights from Non-Dystrophic Myotonia (NDM) and Myotonic Dystrophy (DM) Collaboration

Why Patient Experience Data are so important

Patient experience data (PXD) has become a cornerstone of meaningful patient engagement. Co-creation of such data, carried out jointly by patient experts, industry teams, and facilitation partners, ensures that evidence reflects the real lived experience of those affected, **particularly in rare diseases where traditional clinical datasets are inherently limited**. Health systems increasingly rely on patient-generated insights to guide regulatory, clinical, and access decisions. This is where structured collaboration becomes essential to producing credible, high-impact outcomes.

Understanding Non-Dystrophic Myotonia (NDM) and Myotonic Dystrophy (DM) Through Co-Created Evidence (2017–today)

Since 2017, admedicum has collaborated with Lupin Neurosciences and patient advocates across Europe, the UK, and the United States, to co-create high-quality patient experience data on NDM and DM. This collaboration involved people living with NDM and DM, caregivers, clinical experts, patient organisations, and Lupin's Medical Affairs team, all working together to map disease burden, support treatment access, and document unmet needs in systematic, internationally coordinated projects. Patient experience data, suitable for clinician and regulatory use, were peer-reviewed and presented at international conferences.


RevEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA: preliminary results of the ENSA survey

ensa SURVEY

Introduction/Objectives

Methods

Results

Conclusions

The partnership resulted in several landmark outputs: First and foremost, The European patient survey was an important component for supporting the EMA's decision to maintain the orphan-drug-status of Mexiletine for NDM in November 2018. Following that starting point, further outputs resulted in a burden-of-disease survey, a multi-country study on treatment access barriers, and an international patient-reported survey on the impact of myotonia in DM. These studies, co-created with patient representatives from the earliest design steps through dissemination, have contributed to the advancement of global understanding of NDM and DM.

Perspectives from the Collaborators

"As regulatory and HTA bodies increasingly integrate patient experience into decision making, co creation models are becoming essential in rare neuromuscular diseases. These approaches expand the understanding of disease burden beyond conventional clinical endpoints and ensure that patient experience meaningfully informs regulatory and development decisions."

- Alla Zozulya Weidenfeller, Head Lupin Medical Affairs Neurosciences

"Lived experience isn't just anecdotal - it's essential. When researchers meaningfully engage patients, they gain insights that can fundamentally change how studies are designed, how outcomes are measured, and how successful a therapy can be in everyday life."

- Janet Stone, Epidemiologist (MPH) and Board-Certified Patient Advocate specializing in non-dystrophic myotonia (NDM)

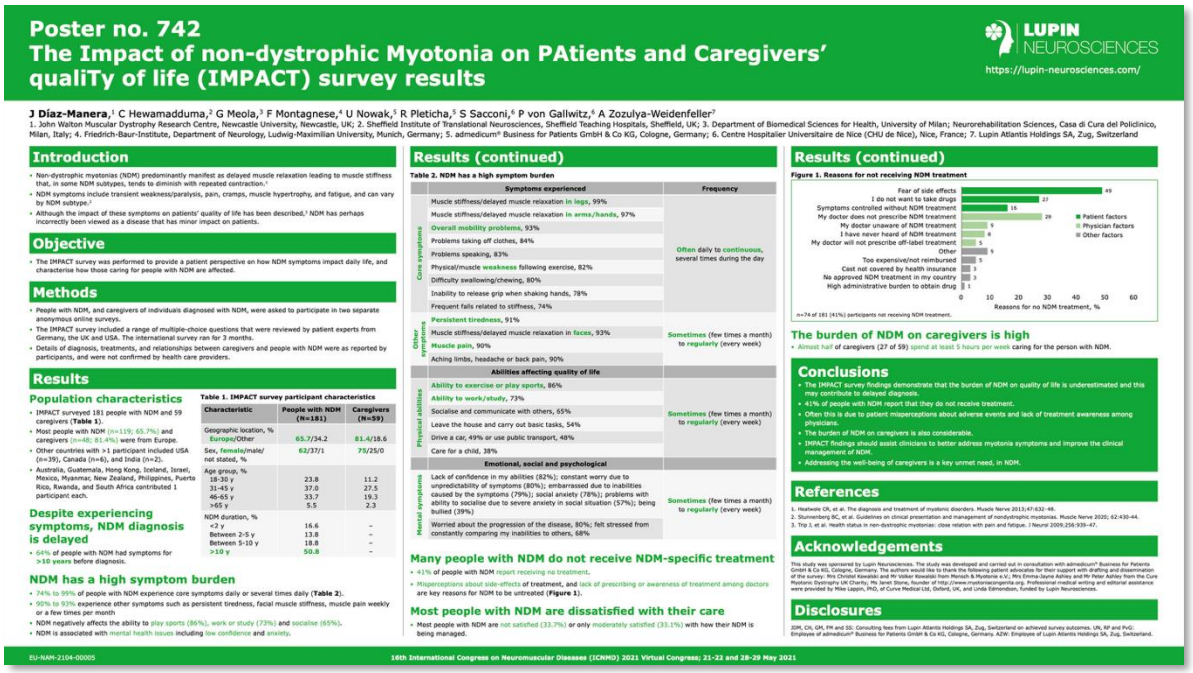
"Complex, rare diseases such as Myotonic Dystrophy are often not fully understood and, because of the familial burden, are subsequently under-represented. Ensuring the patient voice is heard, and lived experience is understood, is essential for making meaningful differences to the community and vital for accurately representing this heterogenous cohort through the drug development and regulatory journey."

"In rare and complex conditions, the lived experience of the patients and their carers is fundamental to ensure that new treatments focus on the symptoms that are actually important to the patients. This work demonstrates that conditions with differing medical causes can benefit from an effective collaboration of organisations looking at the impact of shared symptoms in the real world."

- Emma-Jayne and Peter Ashley, Founding Trustees, Cure Myotonic Dystrophy UK Charity

Exemplifying State-Of-The-Art Patient Experience Data Collection

Across all publications, the collaboration followed a consistent patient-engagement logic: early involvement, equal-footed contribution, methodological transparency, and continuous dialogue. The collaboration ensured that patient representatives were not merely consulted but meaningfully involved in shaping research questions, reviewing survey tools, interpreting results, and validating the narrative used in external communications.



Poster no. 742
The Impact of non-dystrophic Myotonia on PAtients and Caregivers' qualiTy of life (IMPACT) survey results

LUPIN NEUROSCIENCES
https://lupin-neurosciences.com/

J Díaz-Manera,[1] C Hewamadduma,[2] G Meola,[3] F Montagnese,[4] U Nowak,[5] R Pleticha,[5] S Sacconi,[6] P von Gallwitz,[6] A Zozulya-Weidenfeller[7]
1. John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle, UK; 2. Sheffield Institute of Translational Neurosciences, Sheffield Teaching Hospitals, Sheffield, UK; 3. Department of Biomedical Sciences for Health, University of Milan; Neurorehabilitation Sciences, Casa di Cura del Policlinico, Milan, Italy; 4. Friedrich-Baur-Institute, Department of Neurology, Ludwig-Maximilian University, Munich, Germany; 5. admedicum® Business for Patients GmbH & Co KG, Cologne, Germany; 6. Centre Hospitalier Universitaire de Nice (CHU de Nice), Nice, France; 7. Lupin Atlantis Holdings SA, Zug, Switzerland

Introduction

- Non-dystrophic myotonias (NDM) predominantly manifest as delayed muscle relaxation leading to muscle stiffness that, in some NDM subtypes, tends to diminish with repeated contraction.[1]
- NDM symptoms include transient weakness/paralysis, pain, cramps, muscle hypertrophy, and fatigue, and can vary by NDM subtype.[2]
- Although the impact of these symptoms on patients' quality of life has been described,[3] NDM has perhaps incorrectly been viewed as a disease that has minor impact on patients.

Objective

- The IMPACT survey was performed to provide a patient perspective on how NDM symptoms impact daily life, and characterise how those caring for people with NDM are affected.

Methods

- People with NDM, and caregivers of individuals diagnosed with NDM, were asked to participate in two separate anonymous online surveys.
- The IMPACT survey included a range of multiple-choice questions that were reviewed by patient experts from Germany, the UK and USA. The international survey ran for 3 months.
- Details of diagnosis, treatments, and relationships between caregivers and people with NDM were as reported by participants, and were not confirmed by health care providers.

Results

Population characteristics

- IMPACT surveyed 181 people with NDM and 59 caregivers (**Table 1**).
- Most people with NDM (n=119; 65.7%) and caregivers (n=48; 81.4%) were from Europe.
- Other countries with >1 participant included USA (n=39), Canada (n=6), and India (n=2).
- Australia, Guatemala, Hong Kong, Iceland, Israel, Mexico, Myanmar, New Zealand, Philippines, Puerto Rico, Rwanda, and South Africa contributed 1 participant each.

Table 1. IMPACT survey participant characteristics

Characteristic	People with NDM (N=181)	Caregivers (N=59)
Geographic location, % Europe/Other	65.7/34.2	81.4/18.6
Sex, female/male/not stated, %	62/37/1	75/25/0
Age group, %		
18-30 y	23.8	11.2
31-45 y	37.0	27.5
46-65 y	33.7	19.3
>65 y	5.5	2.3
NDM duration, %		
<2 y	16.6	–
Between 2-5 y	13.8	–
Between 5-10 y	18.8	–
>10 y	50.8	–

Despite experiencing symptoms, NDM diagnosis is delayed

- 64% of people with NDM had symptoms for >10 years before diagnosis.

NDM has a high symptom burden

- 74% to 99% of people with NDM experience core symptoms daily or several times daily (**Table 2**).
- 90% to 93% experience other symptoms such as persistent tiredness, facial muscle stiffness, muscle pain weekly or a few times per month
- NDM negatively affects the ability to play sports (86%), work or study (73%) and socialise (65%).
- NDM is associated with mental health issues including low confidence and anxiety.

Results (continued)

Table 2. NDM has a high symptom burden

	Symptoms experienced	Frequency
Core symptoms	Muscle stiffness/delayed muscle relaxation in legs, 99%	Often daily to continuous, several times during the day
	Muscle stiffness/delayed muscle relaxation in arms/hands, 97%	
	Overall mobility problems, 93%	
	Problems taking off clothes, 84%	
	Problems speaking, 83%	
	Physical/muscle weakness following exercise, 82%	
	Difficulty swallowing/chewing, 80%	
	Inability to release grip when shaking hands, 78%	
	Frequent falls related to stiffness, 74%	
Other symptoms	Persistent tiredness, 91%	Sometimes (few times a month) to regularly (every week)
	Muscle stiffness/delayed muscle relaxation in faces, 93%	
	Muscle pain, 90%	
	Aching limbs, headache or back pain, 90%	
Physical abilities	**Abilities affecting quality of life**	Sometimes (few times a month) to regularly (every week)
	Ability to exercise or play sports, 86%	
	Ability to work/study, 73%	
	Socialise and communicate with others, 65%	
	Leave the house and carry out basic tasks, 54%	
	Drive a car, 49% or use public transport, 48%	
	Care for a child, 38%	
Mental symptoms	**Emotional, social and psychological**	Sometimes (few times a month) to regularly (every week)
	Lack of confidence in my abilities (82%); constant worry due to unpredictability of symptoms (80%); embarrassed due to inabilities caused by the symptoms (79%); social anxiety (78%); problems with ability to socialise due to severe anxiety in social situation (57%); being bullied (39%)	
	Worried about the progression of the disease, 80%; felt stressed from constantly comparing my inabilities to others, 68%	

Many people with NDM do not receive NDM-specific treatment

- 41% of people with NDM report receiving no treatment.
- Misperceptions about side-effects of treatment, and lack of prescribing or awareness of treatment among doctors are key reasons for NDM to be untreated (**Figure 1**).

Most people with NDM are dissatisfied with their care

- Most people with NDM are not satisfied (33.7%) or only moderately satisfied (33.1%) with how their NDM is being managed.

Results (continued)

Figure 1. Reasons for not receiving NDM treatment

n=74 of 181 (41%) participants not receiving NDM treatment.

The burden of NDM on caregivers is high

- Almost half of caregivers (27 of 59) spend at least 5 hours per week caring for the person with NDM.

Conclusions

- The IMPACT survey findings demonstrate that the burden of NDM on quality of life is underestimated and this may contribute to delayed diagnosis.
- 41% of people with NDM report that they do not receive treatment.
- Often this is due to patient misperceptions about adverse events and lack of treatment awareness among physicians.
- The burden of NDM on caregivers is also considerable.
- IMPACT findings should assist clinicians to better address myotonia symptoms and improve the clinical management of NDM.
- Addressing the well-being of caregivers is a key unmet need, in NDM.

References

1. Heatwole CR, et al. The diagnosis and treatment of myotonic disorders. Muscle Nerve 2013;47:632–48.
2. Stunnenberg BC, et al. Guidelines on clinical presentation and management of nondystrophic myotonias. Muscle Nerve 2020; 62:430–44.
3. Trip J, et al. Health status in non-dystrophic myotonias: close relation with pain and fatigue. J Neurol 2009;256:939–47.

Acknowledgements

This study was sponsored by Lupin Neurosciences. The study was developed and carried out in consultation with admedicum® Business for Patients GmbH & Co KG, Cologne, Germany. The authors would like to thank the following patient advocates for their support with drafting and dissemination of the survey: Mrs Christel Kowalski and Mr Volker Kowalski from Mensch & Myotonie e.V., Mrs Emma-Jayne Ashley and Mr Peter Ashley from the Cure Myotonic Dystrophy UK Charity; Ms Janet Stone, founder of http://www.myotoniacongenita.org. Professional medical writing and editorial assistance were provided by Mike Lappin, PhD, of Curvo Medical Ltd, Oxford, UK, and Linda Edmondson, funded by Lupin Neurosciences.

Disclosures

JDM, CH, GM, FM and SS: Consulting fees from Lupin Atlantis Holdings SA, Zug, Switzerland on achieved survey outcomes. UN, RP and PvG: Employee of admedicum® Business for Patients GmbH & Co KG, Cologne, Germany. AZW: Employee of Lupin Atlantis Holdings SA, Zug, Switzerland.

SINCE 2016

Why we do what we do at admedicum

This approach reflects admedicum's core mission: enabling structured co-creation between patient communities and life-science partners to generate insights that are both scientifically robust and directly relevant to people's daily lives. We feel honored and proud to being part of this strong collaboration!

Contact us!

admedicum® GmbH & Co KG
info@admedicum.com
+49 2236 94733 60
www.admedicum.com

10 years admedicum
PATIENT DRIVEN SOLUTIONS