Health Insurance Profile and Literacy in Myotonic Dystrophy Type 1 (DM1)

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Background

Myotonic Dystrophy Type 1 (DM1)

- DM1 is a complex, rare, progressive neuromuscular disorder driven by mis-splicing of key genes (i.e., a spliceopathy) resulting in multisystem clinical manifestations including skeletal, smooth, and cardiac muscle as well as the central nervous system (CNS) (1,2).
- Individuals with DM1 experience significant morbidity and early mortality, primarily attributed to respiratory or cardiac complications, along with elevated risk of certain cancers (3).
- Consensus care guidelines developed by the Myotonic Dystrophy Foundation and expert physicians recommend regular multidisciplinary involvement in care (4).
- As no disease-modifying therapies are available, DM1 treatment currently consists of symptom management, requiring coordination of multiple medical specialists (5).
- Interest in treatments that influence muscle and CNS symptoms exists and the pace of their development increase the chance of additional options in the future (5).
- To optimize access to new therapies, the DM1 community urgently needs to understand healthcare systems to navigate specialists, medications, and assistive devices for progressive DM1 symptoms.

Health Insurance Literacy (HIL)

- Difficulty obtaining a diagnosis, few healthcare providers familiar with rare diseases, the lack of treatment options, and limited information on natural history often result in the patient or family becoming the disease expert and care coordinator (6).
- Although rare diseases are often multisystemic, care is rarely received in a comprehensive manner, and affected individuals often encounter healthcare silos. Due to a lack of understanding, providers may block access to needed care or services (7,8).
- An association has been shown between low HIL and avoidance of healthcare services due to costs (9,10), which can lead to irreversible progression of rare diseases.
- It is important that individuals with DM1 have sufficient HIL to navigate the healthcare system, choose coverage, understand their benefits, utilize resources covered, and access needed care.

Research Objectives

- To understand the health insurance profile, HIL, and burdens of access experienced by US individuals diagnosed with DM1, comparing HIL to a historical assessment of HIL in the general US population.
- To provide a real-world view of the experience of those in the US living with DM1 regarding burdens accessing care/services, treatments/medications, and devices/equipment for DM1.

Methods

- The study recruited 100 individuals with DM1 or their caregivers through the Myotonic Dystrophy Foundation (MDF), the National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members (University of Rochester Medical Center), and Engage Health's EnCompass® database.
- Participants completed a survey that included demographic information and details used to develop their health insurance profile. HIL was assessed through completion of a quiz previously used by the Kaiser Family Foundation (KFF) to assess HIL in the general US population as the baseline (Figure 1) (11) in order to analyze by insurance management status.
- Participants completed interviews to identify and prioritize access burdens, allocating 100 points across their top three burdens to indicate relative impact. Qualitative data were analyzed in MAXQDA 2020 (VERBI Software, 2019) to uncover both evidence and context for access burdens.
- This mixed-methods, non-interventional study was approved by WCG IRB and conducted in the United States between February and mid-April 2025.

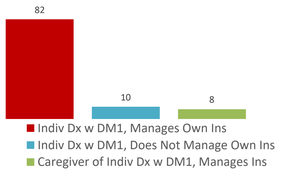
Figure 1. Questions Summary from Norten et al (11). Americans' Familiarity with Health Insurance Terms and Concepts

***Access QR code to review full question*

Q1. Definition, health insurance premium
Q2. Payment of health insurance premium
Q3. Definition, health insurance deductible
Q4. Copay calculation for in hospital stay
Q5. Definition, annual out-of-pocket limit
Q6. Definition, health insurance formulary
Q7. Definition, provider network
Q8. Question re: in-network providers for hospital stay
Q9. Out-of-pocket calculation for out of network lab tests
Q10. Question re: appeals for claim denials

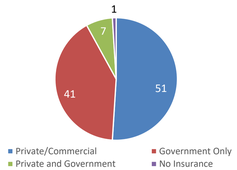
Demographics & Health Insurance Profile

Figure 2: Research Participants, n=100



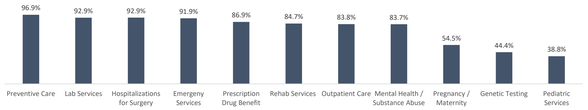
- Of the caregivers who managed insurance (8%), most were mothers- 87.5%.
- 55% of diagnosed individuals were male.
- There were 36 states represented (64%) .

Figure 3: Insurance Plans, n=100



- The insurance plan type proved confusing to participants; based on the expert opinion of an insurance consultant, 23% of insurance type responses were corrected.
- Participants with government only coverage (n=41)
  - 61.0% were on Medicare, 17.1% on Medicaid, 14.6% on Medicare and Medicaid, 4.9% on Medicare and Military insurance, and 2.4% on Military insurance only (including VA).

Figure 4: Health Insurance Covered Services, n=99

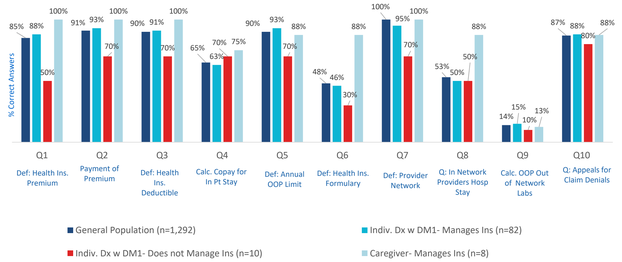


- Most (46.5%) study participants did not require a referral to see a specialist.
- Among the 66 participants who reported their highest individual co-pay for a supportive medication, the mean was $92.53 (range $0–$1,500); 93% reported not participating in any co-pay support, reduced co-pay, or free drug program.

Health Insurance Literacy (HIL)

- When compared with the historical results related to HIL of the general population[11], the DM1 respondents scored better than the general population on 90% of the questions (Figure 5).
- The DM1 population scored best (90% or above) on questions related to *defining a provider network, how premiums are paid, defining health insurance deductible* and *annual out of pocket limit*.
- The DM1 group and the general public scored poorly (below 60%) on questions that asked them to *calculate out-of-pocket expenses, define a health insurance formulary* and *calculate co-pay for an in-patient stay*.
- Within the DM1 population, caregivers managing insurance had the highest percentage correct (70%).
- One participant with DM1 lacked current insurance coverage and was categorized as "does not manage insurance" throughout the study.

Figure 5: HIL of DM1 Cohort (n=100) vs. General Population (n=1,292)



Burden of Access

Figure 6: Relative Burden of Access in DM1 Across All Participants, n=100

Burden of Access Theme	Weighted Score	Burden of Access Theme	Weighted Score
Process to get; care and services	31.89	Process to get; devices and equipment	2.8
Insufficient coverage; care and services	9.33	N/A	2
Not covered; care and services	8.54	Process to get; general coverage	1.95
Time I spend; care and services	8.45	Delay in getting; devices and equipment	1.78
Process to get; treatments and medications	6.65	Delay in getting; treatments and medications	1.75
Delay in getting; care and services	5.95	Insufficient coverage; devices and equipment	1.38
Insufficient coverage; general coverage	5.3	Insufficient coverage; treatments and medications	1.31
Not covered; devices and equipment	2.9	Fear; general coverage	1.15
Not covered; treatments and medications	2.89	General knowledge; resources	1.05
Fear; treatments and medications	2.83	Fear; care and services	0.1

- In this study, the key issue for most participants was the difficulty navigating the healthcare system to obtain care and services ("*process to get*", "*insufficient coverage*", and "*not covered*"), with *process to get care and services* as the most important of all.
- Many participants specifically cited the challenge of finding knowledgeable healthcare providers.
- This theme remained the most important when considered by a number of variables; including individuals diagnosed with DM1, caregivers, those managing their own insurance, and those whose insurance is managed by someone else.

Summary

- This research highlighted the health insurance profile and HIL in a real-world population of individuals diagnosed with DM1 whose symptoms began at age 11 years or later.
- Nearly all (99%) individuals were insured. However, 25% were confused about their insurance plan type, which can result in delayed or denied care, financial strain, and insufficient understanding of plan benefits.
- Caregivers who manage insurance for their loved one (n=8), scored higher on 7 HIL categories compared to individuals living with DM1 who managed their own insurance.
- The study highlighted finding knowledgeable care as a significant burden for the DM1 community.
- The *process to get care* was a prominent issue for participants, and without adequate support, the introduction of novel therapeutics or clinical trials could add additional burden or confusion.

Recommendations

- While most participants were covered by insurance, gaps remain in understanding and navigating health insurance to optimize access in this progressive condition.
- Education to improve HIL is needed, especially for those transitioning to independent coverage, with consideration for cognitive impacts of DM1 in order for maximal benefit to be obtained by participants.
- Key knowledge gaps include how to calculate out-of-pocket costs, how to manage appeals, and the role of drug formularies, which will be important as new therapies become available in DM1.
- Resources to identify DM1-experienced clinicians and select appropriate insurance plans may improve care access and coverage alignment.
- Ongoing practical support is needed to help individuals access and use insurance as new therapies emerge.
- With new medications on the approval horizon, education on patient assistance programs, commonly prescribed medications, and prior authorization will be increasingly important.
- Expanding awareness and use of Medicaid waivers and financial assistance programs may reduce burden amid high costs and reduced workforce participation.
- Efforts to expand coverage for services such as genetic testing (reported as the least covered) may:
  - Reduce long-term care costs
  - Support earlier and more accurate diagnoses
  - Be a pre-requisite for payors in the prescribing of novel therapies in the US

Study Limitations

- Potential limitations of the study may include a recruitment bias in that many were drawn from a population connected to a prominent advocacy organization and may represent individuals who are more informed about their disease.
- The relatively small sample of caregivers managing insurance may also limit select findings.

Acknowledgements

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Disclosures

The research was sponsored by Dyne Therapeutics and conducted in conjunction with the MDF. Engage Health drafted the survey and interview guide in conjunction with Dyne and the MDF, fielded the study and performed all analyses. Interviews were conducted by Engage Health personnel who were trained in the collection of qualitative information. The DM1-Activ^C and the MAASTRICHT UMC+ "DM1-Activ^C Calculation Tables" were licensed from prof. dr. I.S.J. Merkies and Prof. Dr. C.G. Faber, from the School for Mental Health and Neuro Sciences MHeNS, part of the Maastricht University. The MDHI was developed by Heatwole and colleagues and was licensed from the University of Rochester, Rochester, New York. Deidentified patient-level data was provided to Staff from the University of Rochester, who performed all calculations related to the MDHI data. MAXQDA 2020 was utilized through a license obtained from VERBI Software, GmbH. The insurance plan type was reviewed and categorized by Jennifer Shumsky of JLS Consulting, based on the plan name. AD and KB were employees of Dyne Therapeutics, Inc. at the time the study was conducted.

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