Patient-Reported Experiences and Preferences Regarding Palliative and End-of-Life Care in Myotonic Dystrophy: A UK Community Survey

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Introduction

Myotonic Dystrophy Type 1 (DM1) (OMIM#160900) and Type 2 (DM2) (OMIM#602668) are progressive, currently incurable, genetic disorders with multisystem involvement, and reduced life expectancy[1]. DM presents with muscle weakness, myotonia, cardiac conduction abnormalities, respiratory compromise, and other systemic manifestations, yielding complex care needs and increasing morbidity over time. Despite the progressive nature of the condition, palliative care services and Advance Care Planning (ACP) remain underutilised, with delayed referrals limiting comprehensive supportive care and potentially contributing to increased hospitalisations and suboptimal end-of-life experiences[2].

Congenital DM1 (CDM1) is the most severe subtype of DM1, characterised by symptoms apparent at birth or within the first month of life. Childhood onset DM1 (ChDM1) presents with symptoms evident between 1 month and 18 years of age. Previous CureDM surveys identified a marked difference in patient experience, support received, and disease progression for those with symptoms before puberty. Palliative and end of life care is poorly understood in this cohort.

This Charity survey aims to understand illnesses and hospitalisations among individuals affected by DM within the UK, along with patient reported measures of symptom severity and impact, from both patient and caregiver perspectives. Understanding the lived experience, service utilisation, and preferences relating to palliative care and ACP is crucial in enabling better support for patients. Deeper understanding into the burden of illnesses will support the development of emerging therapies, as regulatory bodies are placing increasing importance on the use of real-world evidence in their assessment of new drug applications.

Methods

In March 2026, CureDM published an anonymised online questionnaire designed to capture in-depth data on the epidemiology and patient preferences of the UK DM population. This ongoing survey asked questions about experiences over the last 12 months and was distributed to DM patients and their families via **CureDM UK Charity** networks and the **UK DM Patient Registry**. Respondents included DM patients or their caregivers, with provision to share experiences from the last 12 months of life for loved ones who had passed away.

Data collected included demographic variables (year of birth, sex, residential location), clinical history (age at onset and diagnosis, recent illnesses, hospitalisations), and experiences with palliative services and ACP. Responses were analysed to identify patterns of service engagement and unmet needs.

The primary focus of this survey was to identify recurring causes of illness and hospitalisations, to potentially identify when to involve palliative services, and when to introduce the idea of ACP. The aim is to understand the impact of disease progression and improve care/quality of life for patients and caregivers.

Results

1. Demographics: 212 discrete responses were collected from the UK.

Questions asked age of symptom onset and were categorised as follows: 30 Congenital DM1 (birth to 1 month), 18 Early onset ChDM1 (1 month to 9 years), 35 Late onset ChDM1 (10 to 17 years), 72 Adult-onset (18 to 39 years) and 39 Late adult onset (40 to 55, and 55+). To allow for future analysis, data were gathered for 3 age ranges within adult onset and 2 within late adult and combined for the purpose of this poster. This study also included 18 people living with DM Type-2 The distribution of age within each disease subtype can be seen in **Figure 2**.

Fig 1. Familial disease inheritance data

Disease inheritance data revealed the majority of CDM cases were a consequence of maternal inheritance, however 10% of cases were due to paternal inheritance. Childhood and adult onset favoured paternal inheritance, with late onset and DM2 showing no notable bias. **Figure 1**.

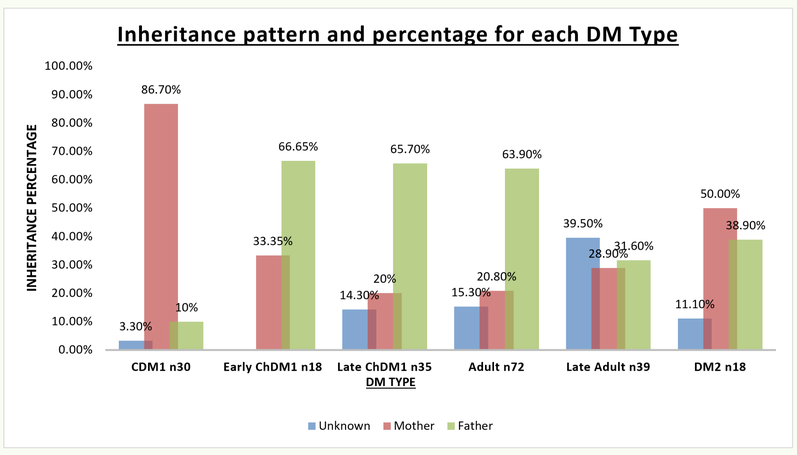
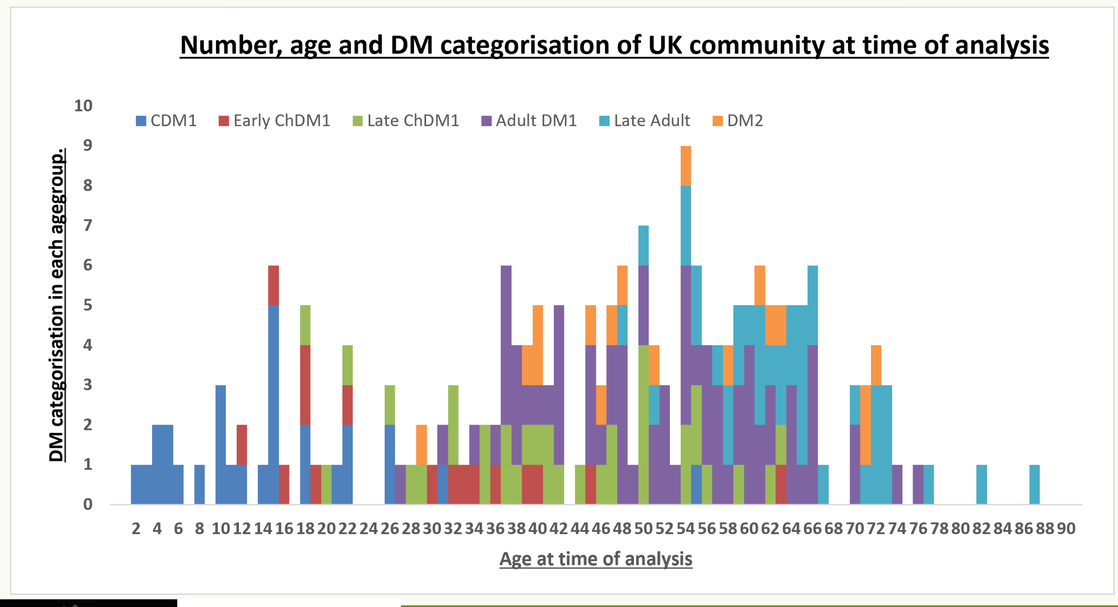


Fig 2. Age distribution by disease subtype in the patient cohort



2. Patient Reported Illness and Hospitalisations.

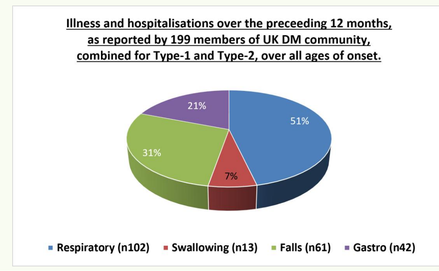
Table 1 shows the percentage of illnesses treated at home, along with combined emergency visits and hospitalisations experienced by each subgroup over the last 12 months. Individuals reported some hospitalisations lasting many weeks, and a snowball effect of an initial cause leading to further systemic involvements. Over the whole cohort, a mean of 59% reported at least one illness over the last 12 months.

Table 1: Illnesses at home, and hospital visits over the last 12 months, for people who answered these questions.

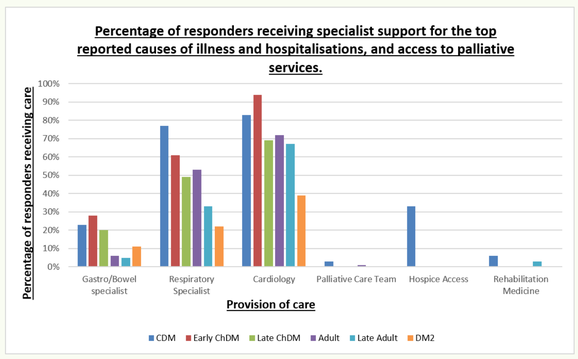
Home	CDM (n27)	Early ChDM (n18)	Late ChDM (n27)	Adult (n70)	Late Adult (n37)	DM2 (n18)	Hospital	CDM (n26)	Early ChDM (n18)	Late ChDM (n27)	Adult (n71)	Late adult (n37)	DM2 (n18)
None	48%	89%	59%	54%	86%	50%	None	59%	83%	74%	72%	76%	70%
1 to 4	41%	11%	37%	43%	14%	22%	1 to 4	39%	9%	24%	27%	24%	22%
5+	11%		4%	3%		28%	5+	2%	9%	2%	1%		3%

As seen in **Figure 3**, causes relating to respiratory, gastro and falls are most reported across all subtypes, with 218 discrete events reported from 199 individuals. Notably, the number of falls was seen to be higher in later onset of disease and DM2, with respiratory being the highest cause of morbidity for CDM.

Fig 3: Most common reported cause of morbidity in DM



3. Patient Access to specialist care.



Caregivers frequently reported avoidable adverse circumstances and gaps in coordinated care, particularly related to gastroenterology. Surprisingly few responses included cardiac involvement, possibly due to cardiac events often being asymptomatic, or the high level of specialist cardiologist involvement.

Figure 4 depicts the minimal input received from specialists, despite the significant number of events.

Analysis identified an almost complete absence of palliative care or hospice support for the whole community, with only CDM reporting service utilisation.

Fig 4: Patient access to specialist services for both palliative support and the top reported causes of morbidity.

Figures 5 and 6 show that 93% and 96% of responders state they never been spoken to within their clinical appointments about palliative care or ACP.

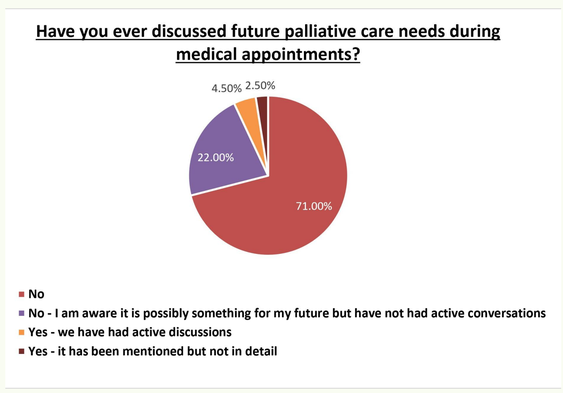


Fig 5. Patient experience of discussing Palliative care in clinic.

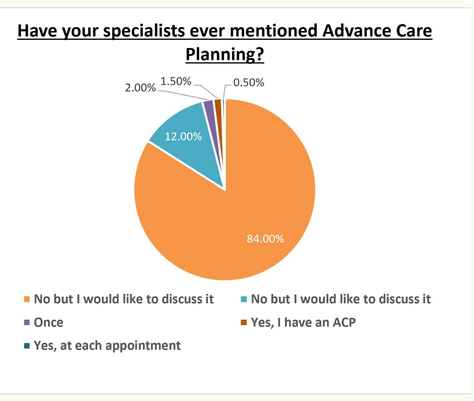


Fig 6. Patient experience of discussing ACP in clinic.

Conclusions

People reporting focused support via home respiratory services described fewer unplanned hospitalisations and greater access to hospice support, yet referrals to palliative care clinics were greatly lacking. Family members of deceased loved ones noted that discussions on end-of-life care were initiated “too late,” with deterioration signs frequently overlooked or underplayed, resulting in emergency hospital admissions and deaths in hospital settings rather than patients’ preferred locations.

The survey reveals an almost complete absence of palliative care for all but a small number of people living with CDM, despite the significant deterioration in health reported. There is significant need for early integration of palliative care and structured ACP in DM management. Charities and support groups are trusted conduits for collecting patient-reported data, offering vital insights into unmet needs. These findings underscore the importance of improving palliative care pathways to enhance patient-centred care and inform clinical trial design across the disease spectrum.

CureDM hope these survey findings will be used to facilitate improved patient experiences within healthcare and break down the stigma around PEOLC. The implementation of an ACP can improve quality of life, make wishes and preferences known, and help keep patients out of hospital by recognising and attending to illness early

References

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[2]Willis D, Willis T, Bassie C, Eglon G, Ashley EJ, Turner C. Myotonic dystrophy type 1: palliative care guidelines. BMJ Support Palliat Care. 2024 Jan 22. DOI: 10.1136/spcare-2023-004748

Acknowledgements and Contact

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