

## **Patient reported illnesses and hospital admissions for Congenital and Childhood onset Myotonic Dystrophy Type 1 over a 12month period.**

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Congenital myotonic dystrophy (CDM) is characterised by symptoms apparent at birth or within the first month of life. Childhood onset DM1 (ChDM) presents between 1-month and 18 years of age, with more severe disability seen in those symptomatic before puberty / 10 years old. CDM and pre-pubescent ChDM often present with complex medical needs, exacerbated by symptoms of Autism, ADHD and learning disabilities.

This study was compiled by CureDM (UK Charity) and presents patient-reported data from the UK myotonic dystrophy (DM) community.

The aim is to provide up-to-date evidence on the burden associated with illnesses and hospital admissions for paediatric patients with DM1 in the UK. An anonymised online questionnaire was sent to DM patients in the UK via CureDM UK Charity and the UK DM Patient Registry. Responses were reported by patients or their caregivers. This paper discusses the responses to the two questions: *‘Have you had any illnesses requiring treatment in the last year?’* and *‘Have you had any hospital admissions in the last 12 months?’*

### **Data Cleaning and Sample Selection**

The original dataset included 427 responses.

### **Filtering Process**

- Excluded 'rest of world' entries: 403 responses remain.
- Removed those without consent: 401.
- Excluded self/carer-assessed ‘adult onset’: 215.
- Re-added those with symptoms before age 10: 219.
- Removed ‘late-adult onset’: 198.
- Excluded DM2 cases: 176.
- Removed self/carer-assessed CDM with symptoms after age 10: 153.
- Excluded self/carer-assessed childhood onset: 107.
- Re-added those with symptoms before age 10: 127.
- Removed duplicates, incomplete, and deceased cases (Total 52).

**75 responses were used for the analysis below.**

### Summary of responses for Congenital DM1 (n=54)

CDM n=54	No	1-4	5-7	8-10	10+
Have you had any illnesses requiring treatment in the last year?	23 42.6%	26 48.1%	2 3.7%	3 5.6%	0
		<b>1 or more 57.4%</b>			
Have you had any hospital admissions in the last 12 months?	32 59%	18 33.3%	2 3.7%	1 1.86%	1 1.86%
		<b>1 or more 40.7%</b>			

### Summary of symptoms/disorders requiring treatment - CDM

Organ class			Number of events
Respiratory disorders	Respiratory tract infections	Bacterial infections	13
		Viral infections	8
Gastrointestinal disorders	Gastrointestinal signs and symptoms	Constipation/impaction	3
		Diarrhoea	1
		Nausea, Vomiting	1
		Stomach cramps	1
Neuropsychiatric events	Convulsion		1

### Summary of symptoms/disorders requiring hospital admission - CDM

Organ class			Number of events
Respiratory disorders	Respiratory tract infections	Bacterial infections	7
		Viral infections	5
	Mechanical ventilation	Neonatal respiratory distress	1
Vascular disorders	Thrombotic and embolic events	Venous thromboembolic event	1
Gastrointestinal disorders	Gastrointestinal signs and symptoms	Constipation/impaction	4
		Diarrhoea	
		Nausea, Vomiting	1
		Bacterial infection	1
Neuropsychiatric events	Convulsion		2

Injury, poisoning and procedural complications	Joint dislocation	Knee dislocation	1
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#### Summary of responses for Childhood onset DM1 (n=21)

ChDM n=21	No	1-4	5-7	8-10	10+
Have you had any illnesses requiring treatment in the last year?	11 52.4%	9 42.9%	1 4.8%	0	0
		<b>1 or more 47.6%</b>			
Have you had any hospital admissions in the last 12 months?	17 81%	4 19%	0	0	0
		<b>1 or more 19%</b>			

#### Summary of symptoms/disorders requiring treatment - ChDM

Organ class			Number of events
Respiratory disorders	Respiratory tract infections	Bacterial infections	2
		Viral infections	3
Renal and urinary disorders	Urinary tract signs and symptoms	Nephrolithiasis	1
		Urinary tract infection	1
Musculoskeletal and connective tissue disorders	Musculoskeletal pain	Back pain	1
General disorders	Cardiac disorders	Cardiac failure	1

#### Summary of symptoms/disorders requiring hospital admission - ChDM

Organ class			Number of events
Respiratory disorders	Respiratory tract infections	Bacterial infections	1
Musculoskeletal and connective tissue disorders	Musculoskeletal pain	Back pain	1
General disorders	Cardiac disorders	Cardiac failure	1
Regular Neuromuscular monitoring as in-patient			1

Support groups and charities have a wide access to patients, often building trusting personal relationships which supports the gathering of comprehensive patient reported data. This firsthand information can be used for wider understanding and for targeting the unmet needs of the community. These data indicate that both congenital and early childhood onset DM come with significant burden for both patients and caregivers in regard to illnesses, with 57.4% (CDM) and 47.6% (ChDM) of responders stating they had required treatment for illnesses within the last 12 months.

There were fewer instances of hospitalisations between the 2 groups, with a significant 40.7% of CDM patients needing hospital care within the last 12 months compared to 19% of childhood onset patients. However, this could partly be due to the reported positive impact of 'rapid respiratory home responders' providing emergency care at home for several children in this cohort who responded as not needing hospitalisations.

Understanding the burden associated with regular illnesses and hospitalisations experienced by children with CDM and ChDM is vital when assessing efficacy of potential treatments. By understanding the risks associated with the disease, there can be a targeted focus for medications to improve the health and wellbeing of patients, thus reducing life threatening situations and improving quality of life for not only the patient, but for the wider family and caregivers. Reducing illnesses and hospitalisations would also reduce the strain on medical services, which currently sees over 50% of CDM children needing regular acute medical support and treatment throughout the year, on top of considerable support already received for their DM1 disease symptoms. Development of treatments for CDM and ChDM is vital to improve the lives of the community. Findings from this survey can be used to enable healthcare professionals and industry to facilitate the planning, design and recruitment of clinical trials. The results also provide a focus on current unmet needs and areas requiring improvement in the care and ongoing support for the UK-DM community.

We extend our sincere gratitude to the UK DM community for sharing their valuable experiences and contributing to the development of this information.

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